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Contents

Why did the Welsh Labour 'Free Home Care' pledge fail to become a reality? Elite perspectives on policy failure.

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Abstract

This paper outlines the prominence of home care for disabled people in the UK and Wales before introducing the debate surrounding paying for such services. It will then give an overview of policy developments in this area since 1997, including the Royal Commission on Long Term Care (Sutherland, 1999), the introduction of free personal care in Scotland following devolution, in 2001 and the failure of the Welsh Assembly Government (WAG) to introduce free home care for disabled people following their 2003 election pledge to introduce such a policy. The research used semi-structured elite interviews with eight elites and one service user. Interviewees' accounts were verified by the use of official documents. It will be suggested that the major factor to have influenced the failure of the policy in Wales was resources. Two other contributory factors were found: firstly, constraints upon the WAG by Local Authorities and the Department of Work and Pensions alongside weak campaigns by the disability rights sector; secondly that there was a change of priorities by the WAG. The article ends with three areas of questioning; should home care be free for all? Did a similar policy succeed in Scotland? And what is the political price of failure? It will be concluded that without the backing of Whitehall, FHC was unlikely to succeed.

Introduction

Within England and Wales almost 20% of the population classify themselves as having a disability that limits their daily activities or work, with one in 16 people of working age being economically inactive as a result of their disability (National Statistics, 2001). The link between disability and poverty is well established (see for example Department of Work and Pensions, 2006). Despite the prominence of disability within the UK, the area of home care receives little attention in research. Furthermore, there is even less information available regarding Wales (United Kingdom Home Care Association, 2007). However, it can be seen that in 2006 almost 25,000 people in Wales were in receipt of financial support for their homecare, (Welsh Assembly Government, 2007a) and that more than 46,000 people over the age of 65 received home care services from their Local Authority (Local Government Data Unit, 2007). It should be noted, however, that this will not include all care, as of the 25,000 people receiving financial support, less than half received Local Authority Care (Welsh Assembly Government, 2007a) therefore the minimum number of people accessing home care services in Wales stands at approximately 58,000 people. This figure excludes all people who use private care providers *and* do not receive financial support for their homecare. The average gross cost of such care has been costed at £13 per hour in 2004/05 (Wanless, 2006) although it is important to note that there is huge variation in charging within Welsh Local Authorities.

Home care for some disabled people is not a luxury item, a point well illustrated by this quotation from a user:

'I can't do without that service, no matter what happens. You know, when push comes to shove I've got to go to the toilet.' (Chetwynd and Ritchie, 1996).

Put simply, without home care, some disabled people would not be able to continue to live in their own home and be independent (Leonard Cheshire, 2007).

Despite the essential nature of such services, many home care users have to pay for their care. Research by the Joseph Rowntree Foundation found that often service users feel that financial assessments do not consider their ability to pay. This has two negative effects: spending has to be curtailed in some other area, including food and heating, which may exacerbate medical conditions (Kober, 2005; Leonard Cheshire Foundation, 2007). Secondly, considerable anxiety could be seen in some recipients regarding the possibility that their care needs, or the price of

their current care, could increase in the future (Chetwynd and Ritchie, 1996). Furthermore, those who may be entitled to some support can be confused and intimidated by the process of applying for an assessment (Leonard Cheshire Foundation, 2007). With regards to service users who were aged over 70, Demming and King (2002) found that many people who could not afford their home care relied instead upon informal care from friends and family. This finding is supported by the Leonard Cheshire Foundation (2007). It appears unlikely that tomorrow's service users will find paying for their care less of a burden: Deeming and King's (2002) research also examined the incomes of people aged over 50 years and found that they were highly unlikely to be able to afford to pay for home care services later in their lives. However, if one considers the income of today's older people relative to older people prior to the 1940s, it can be seen that they have increased assets in the form of occupational pensions and assets such as property. Whilst it can be seen that paying for home care is difficult for some people, and it is considered likely to remain that way in the future by some research, an important question to ask is should such care be paid for out of public funds for everyone?

Debates surrounding this question have been pushed to the fore in recent years following the Royal Commission on Long Term Care (Sutherland, 1999) that suggested the current system was 'characterised by complexity and unfairness' (4.1). This refers to the variation in charges for services between Local Authorities; the maximum charges by Local Authority in Wales ranged from £16.20 to £185 per week in 2007 (Welsh Assembly Government, 2007b). Moreover Sutherland (1999) suggests that this system 'leads to the impoverishment of people with moderate assets before they get any help' (Chapter 1).

Policy Context

It is not the intention of this paper to provide a description of developments in the funding of home care for disabled people before 1999 (for a good description of developments in the 1940s see Fraser (2003:240-253); for later developments see Timmins, 2001).

Shortly after coming to power in 1997, the New Labour government set up the Royal Commission on Long Term Care. The Commission had two major recommendations within the Majority Report (Sutherland, 1999). These were the introduction of a National Care Commission and that

'The costs of long-term care should be split between living costs, housing costs and personal care. Personal care should be available after assessment according to need and paid for from general taxation: the rest should be subject to a payment according to means.' (Sutherland, 1999:2).

Although the Commission's terms of reference did not include younger disabled people, it was felt that the above recommendation should also be extended to this group. In fact, it was suggested that it might be more important to provide free personal care for working age disabled people, as their likelihood of disablement was seen as more 'random' (9.14) and could potentially have more negative effects. Such an extension was also justified on grounds of equity.

On the other hand, a letter of dissent was presented by two members of the Commission (Joffe and Lipsey, 1999). The Minority Report suggested that in funding all personal care, scarce resources would be removed from the poorest disabled people, and as such the policy recommended by Sutherland (1999) would fail to be redistributive. Moreover, it was suggested that for a number of reasons the cost of care would be likely to rise in the future, making the policy unaffordable. Finally, the dissenters suggested that, as a means test would continue to be in place for 'hotel' charges, the policy would have less impact upon home care recipients. This view was adopted, to some extent, by the UK government who rejected Sutherland's recommendation for free personal care. It was stated that costs would be high, services might not be improved and the least well off would not benefit (Department of Health, 2000).

However, a newly devolved Scotland introduced a policy of free 'personal care' for older people in 2002 to 'end discrimination' for those with degenerative or chronic illnesses (Care Development Group, 2001:10). Based upon data available for the first three years, the policy has been seen as successful in a number of ways by the Health Committee of the Scottish Executive (2006) and a Joseph Rowntree Foundation study (Bell and Bowes, 2006). Conversely, Bell and Bowes (2006; Bowes and Bell, 2007) found that free personal care was more expensive than anticipated and was a disappointment to some service users who had not realized that 'hotel' costs would not be covered by the policy.

Alongside the introduction of free personal care in Scotland, the Welsh Assembly Government began investigating if a similar policy could be feasible in Wales (see Wales NHS, 2001 for more details). One reason behind this was the long-standing variation in charging for home care (see for example Welsh Assembly Government, 2007b) as a result of some Local Authorities disregarding disability-related benefits such as Attendance Allowance and Disability Living

Allowance, whilst others included these as income during means tests. Fairer charging guidance was introduced in Wales in 2002 to attempt to rectify this. It was suggested that Local Authorities should ensure that there charges were 'fair' and operating within social-care objectives (Welsh Assembly Government 2002a), yet it failed to standardise charging for care, and in fact was utilised by several Local Authorities in Wales as a reason to increase charges for care (Disability Wales, 2005).

In their 2003 Election Manifesto, Welsh Labour had ten top pledges, one of which was to 'Scrap Home Care Charges for Disabled People' (Welsh Labour, 2003). On entering power, Task and Finish Working Groups were set up and it looked as though the policy would be implemented. However, in 2006 a Cabinet Statement by the Minister for Health and Social Services announced that an alternative package of benefits for disabled people would be introduced as an alternative to the free home care policy because it '…would have quickly become unsustainable.' (Gibbons, 2006a). It was suggested by Gibbons (2006a) that this was as a result of research for the Welsh Assembly Government by Professor Bell (2006).

On the basis of this policy situation the key research question to be addressed is:

Why did the pledge to 'Scrap Home Care Charges for Disabled People in Wales' fail to become a policy?

Methodology

The research used a multi-method approach of semi-structured interviewing with a purposive sample of knowledgeable elites alongside documentary analysis. All data were then subject to policy analysis.

In addition to concerns surrounding finding a population who had extensive knowledge of the issues around FHC, it was also desirable to 'study up', that is to interview elites as they had the power to implement this policy (see for example Mills, 1963). Furthermore, to create a more complete picture of the situation and reduce the likelihood of systematic error or bias, several different groups were sampled (Rubin and Rubin, 2005; Goldstein, 2003). This was, however, a small population of 'key informants' rather than a more representative sample. Groups from which elite respondents would be contacted include disability rights organisations, opposition politicians, Welsh Labour politicians and Welsh Assembly Government (WAG) officials. It was

not possible, within the time available, to find a sampling frame of service users who were knowledgeable about the policy. In order to sample from the four groups, a purposive approach was adopted. For example, *The Coalition on Charging Cymru* comprised of 15 disability rights groups (see Coalition on Charging Cymru, 2006:14-16) the most prominent five of these, in terms of the documents examined, were asked to participate. With regards the other three groups, all of the population were sampled; all opposition spokespeople for Health and Social Care, both Ministers for Health and Social Care from the Welsh Labour party, all three of the major civil servants and a senior political advisor who had been involved in the project.

The research was carried out with nine individuals. Jane Hutt, Minister for Health and Social Services at the time of the election pledge; Jonathan Morgan (Conservative) and Kirsty Williams (Liberal Democrats), spokespeople for Health and Social Services; Rhian Davies from Disability Wales; Ian Thomas from the Alzheimer's Society Cymru; a senior political advisor to the Labour Party; two civil servants and a service user. For the purposes of data analysis, these will be divided into three groups: Welsh Assembly Government officials, Jane Hutt, the senior political advisor and the two civil servants. Secondly, opposition politicians, Kirsty Williams and Jonathan Morgan. Finally, the disability rights sector, Rhian Davies, Ian Thomas and the service user. Such distinctions were adopted because of the different allegiances of groups; the WAG officials, some Labour, some impartial civil servants were all linked to the policy, likewise the disability rights groups and service user were campaigning for the introduction of the policy. Finally, the opposition politicians did not fall into either of these categories.

Requests for interviews began in November 2006, in some cases with several follow up e-mails. All interviews were carried out during March 2007. Such delays are common when involving elites in research (Lilleker, 2003; Goldstein, 2002). However, a delay in this case resulted in interviews being carried out two months prior to the Welsh Assembly Government elections when those who are involved in politics are likely to be at their busiest. Non-response is a problem for all interview-based research, and is a particular problem when carrying out elite interviews (Berry, 2002). In that light, and taking account of the political situation at the time, the level of response should be considered high. The non-responders were Brian Gibbons, Health and Social Services Minister at the time of the research, Helen Mary Jones, Plaid AM, three of the disability rights groups and one civil servant. In order to ensure the views of these people were reflected, press releases and other documents available as a result of freedom of

information requests were analysed. In addition to the above groups of elites, one service user took part after their details were passed to me by an interviewee.

There were three further issues when carrying out the interviews. Firstly time constraints. As recommended by the literature on interviewing elites, I had requested half an hour (Goldstein, 2002; Lilleker, 2003). I always arrived slightly early but some interviews ran later than scheduled, interviews were often interrupted and three interviews were less than 12 minutes long. A second issue was the context of the interview. This was something that I had little control over; in several cases a colleague from Cardiff University had facilitated access, these interviews occurred in a public area at the Welsh Assembly, in hurried circumstances with members of the public and staff walking past regularly. Whilst these issues may seem out of the ordinary for social science interviewing, when research involves elites, people with busy schedules and high levels of responsibility, they should be considered the norm. It would be inappropriate for a senior political figure - be they an AM, a civil servant or the head of a charity – to suspend their official duties for an interview with a researcher! A final less common issue was keeping respondents focused. I had prepared an aide memoir of key areas to discuss and the majority of interviewees were happy to attempt to answer my questions as best they could. However, two respondents were more interested in telling me what they thought were the positive and negative points of publicly funded home care, despite my attempts to keep them focused on the policy process.

Alongside data from interviews, all policy documents relating to home care available under Freedom of Information requests[1] were analysed to provide support for interviewees' views. This approach is recommended by Lilleker (2003:208) when carrying out elite interviews as a result of the 'severe limitations' of such a methodology in isolation.

Data were manually analysed using a policy analysis approach, which is a combination of discourse analysis and content analysis. John (2000) suggests that policy analysis usually takes into account only one factor, such as economics, when looking at the success or failure of policies, but that this should not be the case and that many aspects should be explored. These are five factors, resources, interrelationships, constraints, norms and interests. Although his approach is not adopted in its entirety, it certainly informed that data analysis.

[1] Individual references for each document are not given here, although they are available on request. The page to search for such documents is http://www.information.wales.gov.uk/content/extrareports/index-e.htm Accessed 30.03.08

Findings

Data from the interviews are analysed in relation to three key themes: resources; constraints and campaigns; and interests. It should be noted, however that the data are complex and dividing them into discrete categories is by no means straightforward. Initially John's (2000) approach was adopted, however this was modified to provide a better fit with the data. Likewise there was considerable variation between different groups of people; WAG officials, opposition politicians and disability rights sector.

Resources

The most common factor discussed was that of resources. This was mentioned by all interviewees and was discussed at greater length than either interests or constraining bodies. In addition, it was the only factor discussed by the opposition politicians. Within this area, two approaches can be seen. Firstly, the idea that the policy would have proven too expensive. The alternative approach suggested that the cost of the policy was affordable, as such the idea that the policy was too expensive was simply used as an excuse to justify not implementing FHC. The latter opinion was found only within the Service User's interview.

This factor was discussed most by the two civil servants who unveiled multiple factors that, in their opinion, were likely to increase costs. These included potential increases in demand for services in the future, the impact of the definition of 'disabled people' on cost and the higher numbers of Attendance Allowance - and other disability related benefit – claimants within Wales as compared to the rest of the UK. No other person or group argued this case so persuasively.

Other respondents, from all three groups of respondents, stated that the WAG 'could not afford' the policy, without giving such detailed insights as to why this might be the case (Jonathan Morgan). However the service user did suggest that:

'I know it's a money issue. I know that after the budget the Welsh Assembly Government has about £15 billion, and politics, to quote Nye Bevan, is the language of priorities.'

Showing that it was a choice for the Assembly to 'not afford' the policy. The civil servants second issue of latent demand was also discussed by other respondents including members of the WAG officials group and the disability rights sector, although it was not mentioned by either

opposition politician. The third issue from the civil servants was also identified by another interviewee: the idea of 'the demographic reality of Wales', that is the higher numbers of claimants of disability related benefits compared to the UK, was identified by Ian Thomas as a particular hurdle for the WAG to overcome. A final factor identified, by all respondents apart from the WAG officials was that of ignorance:

'…I think they'd gone into an election with big policies that they could sell without sitting down and thinking about the consequences of what they were saying. They hadn't really thought about how they were going to pay for what they were saying.' (Kirsty Williams, Opposition Politician.)

Constraints and Campaigns

The second most common theme emerging from the research was that of pressure exerted by organizations outside of the WAG. These factors were mentioned primarily by the WAG officials with the only other mentions being by the Service User. This can be divided into interactions between the WAG and three bodies; Local Authorities, the Department of Work and Pensions (DWP) and disability rights groups. The most discussed of these issues is that of Local Authority reluctance to implement the policy. All WAG officials agreed with this. It was suggested that Local Authorities were concerned about the cost of the policy and about work force issues:

'Very quickly Local Authorities said they weren't happy… because it was removing their discretion and freedom. And you needed not to have this; in terms of primary legislation we had no way to force the Local Authorities to remove the charges.' (Jane Hutt, Minister for Health and Social Services)

Furthermore it was noted that:

'…implementing a policy in the face of opposition from the people who've got to make it work on the ground is pretty hard going.' (Senior Political Advisor.)

These views were not shared by the Service User, who failed to be persuaded that Local Authority opposition was a serious threat to the policy; however, like the Service User's views on resources, they were not mirrored by any other respondent.

Less attention was paid to the constraining influence of the DWP, although it was noted by all WAG officials that there could be problems if the criterion by which a person was defined as 'disabled' was related to a DWP benefit. If this were to be the case Whitehall, rather than Cardiff, could indirectly control who received financial support for their home care. A second issue that was mentioned less frequently was the potential for the DWP to withdraw Attendance Allowance payments which could cost the WAG several million pounds per annum. This had occurred in Scotland following the introduction of Free Personal Care and had accounted for a large increase in costs (Bowes and Bell, 2007).

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The final issue within this factor is that of the voluntary sector. This issue was only raised by the Senior Political Advisor who suggested that the disability rights groups involved had failed to 'fight the fight' in order to secure the centre stage position within the Assembly. Support for this view can be found within the interview with Rhian Davies who describes a lobby at the Welsh Assembly building as having 'quite a good turn out' when fifty people were present, likewise despite template letters being made available by the Coalition on Charging, Rhian Davies stated that Assembly officials had told her that 'the Minister's postbag was light'. An extension of the Senior Political Advisor's view can be seen from the Service User who suggests that had 'ordinary people' supported the campaign more strongly, it would have been more difficult for the WAG to not fulfill their manifesto pledge.

Looking at the interaction between the WAG and these three bodies, and following the Senior Political Advisor's train of thought, there was considerable pressure on the WAG not to implement the policy from both the Local Authorities and the DWP. Such pressure was not equalised by the voluntary sector who were campaigning for the policy.

Interests

The final category is that of interests. This was discussed primarily by the Service User who devoted considerable time to his explanation. It was stated that 'there was a change of emphasis when the Minister (for Health and Social Services) changed' from Jane Hutt to Brian Gibbons as Jane was from 'more of a voluntary sector background' and as such was seen as more of a driving force for the policy.

A second interpretation of this category can be seen from the WAG officials (with the exception of Jane Hutt). In this instance, the policy would undermine the idea of joined-up policy and create a disappointment to service users, due to unavoidable financial constraints, and as such, in the face of other considerable difficulties, it was no longer in the WAG's interests to pursue the policy:

'If we head down this route, we're going to have a mean and narrow definition of what we mean by 'disabled' and a pretty mean and narrow definition of what we mean by 'home care' but we will find definitions of those that are workable on the ground and we will then have a group of people who will benefit from the policy.' (Senior Political Advisor.)

Within all sections, interviewees' accounts were supported by documentary analysis. This is somewhat unusual when carrying out elite interviews (Lilleker, 2003). However, this is not to suggest that interviewees held similar views. Within two of the three groups there was little variation; the WAG officials spoke at length about the difficulties in attempting to introduce the policy as a result of constraints and campaigns. All respondents focused a lot of attention on the area of resources, although the opposition politicians focused upon it exclusively. The one group in which there were significant differences is the disability rights sector where the Service User held significantly different views to all other respondents in several areas. This may be as a result of the differing position of the Service User in terms of not being actively involved in attempting to introduce policy. This might also explain why the WAG officials discussed a wider range of factors than the other groups; put simply they had more experience in attempting to implement the policy.

Discussion

Should home care be free for everybody?

This is of course a normative question which cannot be resolved simply by the evidence. Within the earlier discussion of policies it can be seen that the Majority Report of the Royal Commission on Long Term Care (Sutherland, 1999) and the Scottish Executive (Care Development Group, 2001) along with the disability rights sector (for example, Leonard Cheshire, 2007) believed that home care should be provided free of charge to all disabled people aged over 65. However, the Minority Report (Joffe and Lipsey, 1999) and the UK government (Department of Health, 2000) believed that the cost of such a policy would be unaffordable and that people should provide for

their own care (unless they are eligible for means-tested support). Within the interviews, Jonathan Morgan suggested that people should be expected to support themselves, with all other respondents broadly supporting the idea of the policy. However, this is not to suggest that the other respondents did not identify the difficulty in financing the policy. It was suggested by several respondents that if the policy had been implemented within the original budget, it would be likely to disappoint service users. With reference to the review of the Scottish policy by Bowes and Bell (2007), this notion does appear likely; in Scotland some service users were disappointed as a result of having misunderstood what the policy would cover. If the policy were to have a narrow definition of 'disabled', many service users who consider themselves to be disabled, would be ineligible for free home care.

Did a similar policy succeed in Scotland?

The research question asked why did the pledge to 'Scrap Home Care Charges for Disabled People in Wales' fail to become a policy? However, within interviews, many respondents referred to the Scottish situation. Thus, with hindsight, it may have been more appropriate to extend the question to ask: Why did the pledge to 'Scrap Home Care Charges for Disabled People in Wales' fail to become a policy when the Scottish Free Personal Care policy was implemented? I will briefly outline some relevant factors. The political situation can be seen to be of considerable importance: as part of a coalition government, Henry McLeish[2] was pressured by the Liberal Democrat part of the Executive into making the pledge into a policy reality. This pressure was not present in Wales. Moreover, the policy, as the first major deviation from UK social policy in devolved Scotland, was afforded considerable importance in showing that the Scottish Executive was a capable government. It has been suggested by journalists that had the policy failed, it would have been 'political suicide' for Jack McConnell[3] (Nutt and Farquharson, 2006).

The factor of cost, which was prominent in interviews, has a different impact in Wales compared to Scotland. Initially in Scotland the Free Personal Care policy was estimated to cost an affordable £50 million (Care Development Group, 2001). This is more than double the estimated cost of the Welsh scheme. Furthermore, the concerns of the WAG officials in

[2] Henry McLeish was First Minister for Scotland at the time and was involved in a political scandal surrounding expenses claims, resulting in his resignation soon after the commitment to introduce Free Personal Care in Scotland.
[3] Jack McConnell succeeded Henry McLeish as First Minister of Scotland in 2001.

interviews around losing DWP funding for Attendance Allowance were realized in Scotland, increasing the cost of their policy by 20% (Bowes and Bell, 2007). Whilst this is far from desirable for the Scottish Executive, it can be afforded as a result of Scotland's relatively stronger financial position resulting from the Barnett formula and the Executive's ability to raise taxes in order to create revenue. Such an increase could be disastrous for Wales. It is perhaps for this reason that the Sutherland Report's (1999) recommendations were aimed at administrations which can raise money in addition to spending it.

The political price of failure

Whilst interviewees were not questioned about the political price of failure for the Welsh Labour Party, it was given considerable attention by respondents from all three groups. For example the Senior Political Advisor suggested that opposition parties aimed to make the Labour Party be seen as:

'…a government that doesn't see things through, a government that you can't trust, that you can't rely on…'

Perhaps, as in debates surrounding the potential of the free personal care policy in Scotland to fail and, as such for devolution to be seen as a failure, there was a strong political price for the Labour Party to pay. In the May 2007 WAG election, the Labour Party failed to secure a majority government. It is beyond the scope of this paper to suggest reasons behind such a result, but perhaps some of the political 'mud' stuck to Labour's reputation.

Policy failure and the public

Whilst it can be argued that the FPC policy in Scotland was not a complete success, it did become a reality. The elites interviewed regarding the Welsh FHC proposed many factors that were involved in the failure of the policy, including resources, constraining factors within government and the interests of the WAG. However, perhaps the most relevant issue was one that was paid little attention – campaigns for the policy both by the disability rights sector and the public. It was suggested by the Senior Political Advisor that other 'difficult and costly' policies, such as free school breakfasts for all primary school children in Wales, were implemented because of support from relevant bodies, in this case educationalists. Furthermore, examining the case of the approval to use the drug Herceptin within the NHS in early stage breast cancer, the BBC (BBC News, 2006:1) suggests that 'patient power' was relevant, if not

crucial, in the decision to introduce a costly policy. Therefore, it can be suggested that if the political cost of not introducing the policy, ie: public protests and diminishing political popularity, were seen as more worrying than the financial cost of the policy and more difficult than the resistance from Local Authorities, the policy, whether it was to be successful or not, may have been more likely to have been implemented. Reasons for the lack of public support for the FHC policy have not been examined to date but 'sicknote Britain' scandals reported in the media may have had an impact upon the public's view of 'disabled people'.

Conclusion

This article has outlined the shortage of literature around the issue of home care for disabled people. The literature also shows the necessity of home care for some disabled people and the sacrifices that recipients make if they are not eligible for free home care. However, the arguments in favour of making home care universally free are accompanied by compelling reasons to use caution in designing policy within this area. This can be seen in the policy reality of the Free Personal Care policy in Scotland. The results showed that there were three main areas that respondents felt contributed to the failure of the FHC policy. The most commonly stated was resources, it was suggested simply that the policy was going to be much more costly than had been originally anticipated. This was for a variety of highly plausible factors. However, the second group of issues was around practical issues among other government departments; Local Authority resistance would have made implementing the policy difficult. Finally it was suggested that it was no longer in the WAG's interests to implement the policy, either because of their changed priorities or because there was an awareness that within the budget available, the policy would be a disappointment.

With the difficulties experienced within the Scottish FPC policy and the failure of the Welsh FHC policy, it is reasonable to ask should FHC be free for everyone or should the means test remain? There is no easy answer to this question as the variables involved are so complex. Examining the differences between Scotland and Wales, it seems that as FPC created disappointment among service users in Scotland, FHC would be likely to disappoint Welsh service users. However, if FHC were to be introduced by Whitehall, as opposed to a devolved country, it would have the financial resources to succeed. Although it is impossible to know beyond speculations, had the public been behind the FHC policy, either within Wales in 2003 or the UK as a whole following the 1999 Sutherland Report, as they were behind the case of introducing Herceptin to the NHS for early stage breast cancer patients, it may have been

introduced. This conclusion should be noted by the disability rights sector who have been very influential in influencing policy in the past.

References

BBC (2006). *Herceptin. Was Patient Power Key?* (9.06.06) Available on line: http://news.bbc.co.uk/1/hi/health/5063352.stm. Last Accessed 28.09.08.

Bell, D. (2006). *Estimating the Cost of Free Home Care for Disabled People in Wales.* Cardiff: Welsh Assembly Government.

Bell, D. and Bowes, A. (2006). *Financial Care Models in Scotland and the UK.* York: Joseph Rowntree Foundation.

Berry, J.M. (2002). 'Validity and Reliability Issues in Elite Interviewing'. *Political Science Online:* December 2002.

Bowes, A. and Bell, D. (2007). 'Free Personal Care for Older People in Scotland: Issues and Implications.' *Social Policy and Society* 6(3):435-445.

Care Development Group (2001). *Fair Care For Older People.* Edinburgh: Care Development Group.

Chetwynd, M. and Ritchie, J. (1996) *The Cost of Care: the impact of charging policy on the lives of disabled people.* Bristol: Policy Press in association with the Joseph Rowntree Foundation.

Coalition on Charging Cymru (2006a). *Campaign Pack.* Available online at: http://www.disabilitywales.org/1657.html Accessed: 03.04.2007

Deeming, C. and Keen, J. (2002). 'Paying for Old Age: Can People on Lower Incomes Afford Domiciliary Care Costs?' *Social Policy and Administration*:36(5):465-481.

Department of Health (2000). *The NHS Plan: the Government's response to the Royal Commission on Long Term Care.* London: Department of Health.

Department of Work and Pensions, 2006. *Houses Below Average Income (HBAI) 1994/5-2005/06*. Available on line at: http://www.dwp.gov.uk/asd/hbai/hbai2006/pdf_files/chapters/chapter_3_hbai07.pdf Accessed 02.03.08.

Disability Wales, (2005). 'Where's the Free Home Care?' *Disability Wales News:* Issue 65. Available on line at: http://www.disabilitywales.org/1640.file.did accessed 20.05.07.

Fraser, D. (2003). *The Evolution of the British Welfare State*. Basingstoke: Palgrave Macmillan.

Gibbons, B. (2006a). *Cabinet Statement: Free Home Care for Disabled People; 15.2.06*. Cardiff: WAG.

Goldstein, K. (2002). 'Getting in the door: sampling and completing elite interviews.' *Political Science Online:* December 2002.

Health Committee (2006). *10th Report of 2006 (Session 2): Care Inquiry - Summary of Conclusions and Recommendations*. Available online at: www.scottish.parliament.uk/business/committees/health/reports-06/her06-10-vol01-00.htm. Accessed 10.01.07.

Joffe, J. and Lipsey, D. (1999). *Note of Dissent (Minority Report of Royal Commission on Long Term Care).* Available online at: www.archive.official-documents.co.uk/document/cm41/4192/4192-ap1.htm Accessed 10.01.07.

Kober, C. 2005. *In the Balance: disabled people's experience of debt.* London: Leonard Cheshire Foundation.

Leonard Cheshire Foundation, 2007. *Home Free: A study into home care in Wales.* London: Leonard Cheshire Foundation.

Lilleker, D.G. (2003). 'Interviewing the Political Elite: navigating a potential minefield'. *Politics:* 23(3): 207-214.

Local Government Data Unit, 2007. *Performance Indicators for Social Services: Adult Services National Strategic Indicators 2006-2007*. Available on line at: www.dissemination.dataunitwales.gov.uk/webview/index.jsp Accessed 26.05.07

Mills, C.W. (1956). *The Power Elite.* New York: Oxford University Press.

National Statistics (2001). The *Census in England and Wales.* Available on line at: http://www.statistics.gov.uk/census/default.asp accessed 14.03.08.

Nutt, K. and Farquharson, K. (2006). 'Will we Still Care for Him when He's 65?' *The Sunday Times: 11.06.2006*

Rubin, H.J. and Rubin, I. (2005). *Qualitative interviewing: the art of hearing data.* London: Sage.

Sutherland, S. et al (1999). *With Respect to Old Age: Long term care – rights and responsibilities (the Sutherland Report).* London: HMSO.

Timmins, N. (2001). *The Five Giants; a biography of the welfare state.* London: Harper Collins.

United Kingdom Home Care Association, 2007. *An overview of the UK domiciliary care sector, November 2007(7th Edition).* Available online at: http://www.ukhca.co.uk/pdfs/domiciliarycaresectoroverview.pdf Accessed 02.03.08.

Wales NHS, (2001). *Jane Hutt Welcomes Consultation on Fairer Home Care Charging.* Available on line at: http://www.wales.nhs.uk/pressnotices/w01177.htm Accessed 25.5.07.

Wanless, D. 2006. *Social Care Review: securing good care for older people.* London: King's Fund.

Welsh Assembly Government. (2002a). *Fairer Charging Policies for Home Care and other non-residential Social Services: guidance for Local Authorities. Cardiff: Welsh Assembly Government.* [Available on line at: http://new.wales.gov.uk/docrepos/40382/40382313/40382112/4038211113/403821111/NAFWC28-02-e.pdf?lang=en]

Welsh Assembly Government, 2007a. *Assessments and Social Services for Adults 2006-2007*. Available on line at: http://new.wales.gov.uk/topics/statistics/headlines/health-2007/hdw200709193/?lang=en Accessed 02.03.08.

Welsh Assembly Government, 2007b. *Press Release: Greater Powers for Wales sought to secure fairer deal for users of non-residential care services.* Available on line at: http://wales.gov.uk/news/ThirdAssembly/Health/2007/1821400/?lang=en Accessed 02.03.08.

Welsh Labour (2003). *Working Together for Wales: Welsh Labour's manifesto 2003*. Cardiff: Welsh Labour.

Picture this! An intergenerational case study involving participant-directed visual data production

Dawn Mannay

Abstract

This paper presents a methodology for engaging participants in participatory visual data generation. The paper draws upon data generated by participants from a research project that aimed to explore and represent the everyday experiences of working-class mothers and daughters residing on a peripheral housing estate. A central concern of the project was generating a research environment and methods through which the compelling voices of the participants were foregrounded. As an indigenous researcher, I aspired to suspend my preconceptions of familiar territory and therefore I asked participants to engage with self-directed methods of data production. Data was generated through a mapping technique that involved participants producing picture maps of their homes and of significant sites in their local area. The paper focuses on maps that were completed by one mother and her nine year old daughter. The usefulness of the technique for making the familiar strange when the researchers own experience mirrors that of their participants is explored alongside wider reflections on pictorial mapping as a participatory method.

Introduction

It has been argued that there is an urgent need to make space for working-class understandings of locality, particularly the perspectives of women and children if the 'cardboard cut-out stereotypes of the urban poor' are to be replaced with a more nuanced understanding of the complexities of their lives (Reay and Lucey 2000:425). Class differences, then, are as insidious as ever, therefore, class now needs to be understood not just in terms of economics, education and occupation but also in the realm of place, space and identity.

In agreement with the call for working-class perspectives on space (Reay and Lucey 2000; Skeggs 1997; Walkerdine 2001) the central substantive aim of the project was to gain an understanding of participants' impressions and interpretations of their local environment. The intention was not to establish an overall view of the working-class mother and daughters experience but to give specific individuals the opportunity to share their conceptions of place, space and identity.

The relationship between researcher and participant is fundamental to the collection of rich and reliable data (Pole 2007). Consequently, it was also important to address my position as an indigenous researcher and make a deliberate cognitive effort to question my taken for granted assumptions of that which I had thought familiar. Therefore, I was influenced by previous research that has used participants' artwork to refresh the habitual responses of the researcher and to render the familiar setting more visible (Kaomea 2003). Introducing a visual element to the process of data collection can potentially provide different ways of knowing and understanding. Art, therefore, may be a medium that can overcome the confines of language, open up experience and make the familiar strange.

Offering an opportunity for participants to metaphorically describe their social and physical environments could, then, lead to a more complex understanding of a given phenomenon or culture (Radnofsky 1996). Additionally, previous work has found that the use of self-directed visual images as a research method act as an empowering tool in research enabling participants, especially children, to exercise greater control in the data generation process (Cook and Hess 2007; Dodman 2003; Barker and Weller 2003; Ross 2002).

Mapping exercises have proved valuable in previous studies of children's perceptions of their environment where the incorporation of map-making into the research process was described as an effective technique for engaging participants and increasing their participation (Barker and Weller 2005; Darbyshire *et al* 2005; Morrow 2001; Ross 2005). Mitchell (2006) found that children observed elements of the environment that were often unseen or forgotten by adults and their pictures provided an opportunity for them to share their perspectives. Moreover, the use of drawings was reported as a mechanism to increase respondents' degree of control within the interview encounter so that issues were raised at their own pace rather than at the pace of the researcher (Wilson et al 2007).

In previous research drawing has proven to be much more popular with seven to eleven year olds than with teenagers or adults (Barker and Weller 2003). However, Nossiter and Biberman (1990) suggest that drawing can also focus adults' responses so that they concentrate on the most salient features of their environments. Furthermore, the researchers propose that putting forward the unusual request for creativity acts as a tool to motivate respondents to analyse their worlds. Thus, I employed a participant-directed mapping technique of data collection, which

aimed both to empower the participants and 'to make the familiar strange and interesting again' (Erikson 1986, p.121).

One criticism of the inclusion of visual images in social science research is that such visual materials are under analysed, 'serving as little more than illustrative devices' (Ball and Smith 1992:12). There is a need, therefore, to seek to provide a descriptive and nuanced analysis of films, photographs and drawings. However, although images render the world in visual terms they can never be transparent windows on the world as there is a distinction between vision and visuality.

Vision simply refers to the physiological capabilities of the human eye, whilst visuality accounts for the complex ways in which vision is constructed. Thus, visuality and the overlapping term scopic regime, refer to the ways in which audiences bring their own ways of seeing and other knowledges to bear on an image (Rose 2001). The audience, then, actively make their own meanings from an image. Yet, if the research is interested in the ways in which people assign meanings to pictures the study of images as data whose meaning is intrinsic, is insufficient (Banks 2001).

Images have no inherent or structural association, other than that which is 'the audience is educated to expect by convention' (Banks 2001:10). The reading of visual images then suggests that the message lies within the visual image and that analysis provides the opportunity for the image to speak. On the contrary, both literally and metaphorically it is human beings who speak to one another and the author or artist is an active element at the site of production that can be neglected when a visual image is under consideration (Rose 2001). This may be acceptable where the research is centered on classic art or archive material but where the producers of an image are available for further explanation this needs to be addressed. In order to gain an understanding of the internal narrative of the image, then, it is imperative to acknowledge the role of the image maker.

The notion that the most salient aspect in understanding a visual image is what the maker intended to show is often referred to auteur theory (Rose 2001). Auteur theory emphasises the importance of participants' interpretation for understanding the meaning of their drawings and the approach is required on a practical level because the interpretation of the audience is not necessarily the same as the narrative the image-maker wanted to communicate; indeed it can

often be markedly different (Kearney and Hyle 2004). The practice of asking participants to explain the visual images that they create has become a common feature of social science research (Belin 2005; Darbyshire *et al* 2007; Morrow 2001). Thus, in order to present the everyday experiences of working-class mothers and daughters it was vital that I apply auteur theory to ascertain the participants own reasons for creating visual images, rather than giving my own interpretations and assumptions to the pictures.

Methodology

Places and spaces: the research site

The geographical space that is home to the participants is an area, which saw a rapid expansion after World War II, in a building programme to provide 'homes fit for heroes'. The area has since acquired a reputation that many of the residents are keen to dispel. One of the largest housing estates in Europe, the area has become the epitome of the classically disadvantaged council estate.

The poverty yardsticks applied to the estate include high unemployment; high rates of teenage pregnancy; high numbers of lone-parent families and high take-up of free school meals. The estate, then, shares the characteristics of the type of place that forms the spatial core of disadvantage in Britain today. There is a stigma associated with these estates as 'types of places' inhabited by 'types of people' (Haylett 2003). Through metaphors of 'waste, refuse and rejection' these estates become 'demonised' (Reay 2004).

However, against this background of the uneven educational and occupational playing field stands an interlinking network of friends, family and familiarity. As Williamson (2004:1) comments 'on a sunny day, it doesn't look so bad, for Milltown' and the same can be said of this estate. There is an evident culture of solidarity and loyalty among many of the 30,000 residents, for after all, this place is their world, and this is home.

Tina and Chantelle

As Rawlins (2006) maintains, by considering intergenerational relationships it is possible to gain a greater depth of understanding since one can compare different versions of the same story. Thus, the study was interested in considering the views of both mothers and their daughters. In this paper I will focus on Tina (1), who is thirty years old, and her nine year old daughter

Chantelle who reside in a council owned property on a well known street in the heart of the council estate.

Mapping as a participatory methodology

In order to create the opportunity for Tina and Chantelle to express their own perceptions of place and space mapping methods of data generation were introduced. Tina and Chantelle were asked to draw maps of their social and physical environments focusing on elements within their home, their street and the wider locality. The maps acted to aid understanding of the participants' conceptions of their local area and to establish what the features of significance were at different levels of space and place. Tina and Chantelle both drew a map of the inside of their home and a map of significant sites in their local area (see Figure 1) and Chantelle also provided an illustration of the outside of her home. The maps then formed the basis of an interview where I engaged in a tape-recorded discussion with each participant and they provided explanations of their images.

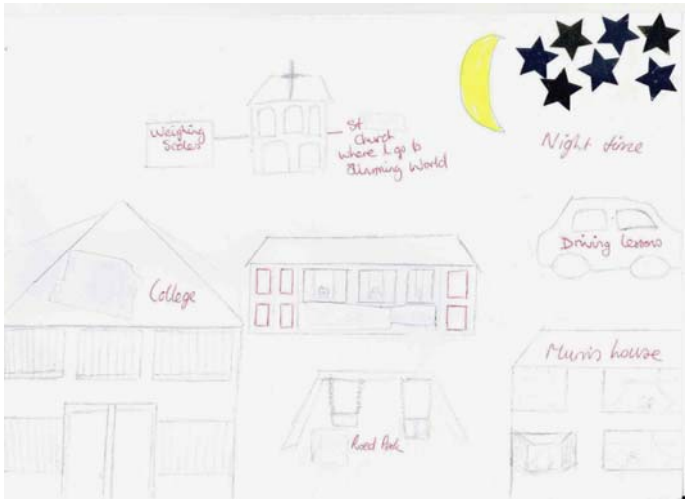

Figure 1: Tina's map of significant sites in her local area

By employing the technique of self-directed visual data collection I was able to gain a more nuanced understanding of the mothers and daughters worlds. Tina and Chantelle provided comprehensive and thoughtful accounts of their lives and their relationship with their immediate locality, offering vivid, eloquent and amusing descriptions alongside candid appraisals of their everyday lives. However, this paper is concerned with the usefulness of the mapping technique within a participatory methodology and its potential for making the familiar strange, rather than the exploration of place, space and identity.

Making the familiar strange

Researchers are often apprehensive about entering a familiar research setting, where their experience with the subject matter, sets up a range of preconceptions about the topic (Vrasidas 2001). As I am a long term resident of the research site, I too was concerned that the findings would be overshadowed by the enclosed, self-contained world of common understanding as the disadvantages of preconceived understandings were experienced in my earlier research, which contributed to an undergraduate dissertation as illustrated below.

'*In the case of the third interview there was a small amount of vacillation because my own children attend the same school as those of the participant. Consequently, there was an element of my questions being unnecessary because I already knew the answers'* (Mannay 2006:21).

There was then, a two-way taken-for-granted cultural competence as I entered the interview with preconceived knowledge and the participant communicated an assumption that I already understood her experience. Thus, conducting research in a culture in which I was habituated had a deadening effect on the interview process in this case. However, although there was a small amount of vacillation in the interview, overall the elements of shared understanding and common ground contributed to a relaxed open atmosphere, which was reflected in the quantity and quality of the data.

Therefore, I decided to remain within the familiar field but to address the taken-for-granted cultural competence inherent to my insider status by employing self-directed visual data collection techniques to promote subject-led dialogue. Tina and Chantelle were not controlled by a pre-determined schedule and they entered the interview setting with their own ideas. This gave me the opportunity to observe and learn about elements of our shared environment and aspects of the participants themselves that I would not have enquired about.

The participant's visual data provided an opportunity to observe unseen or forgotten elements of the physical environment such as Tina's representation of the night sky.

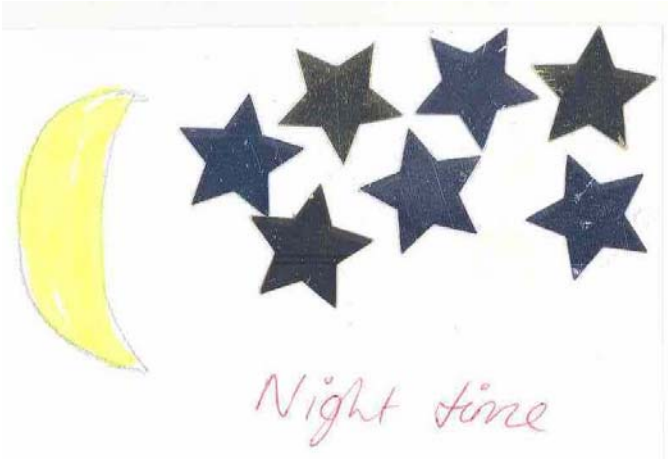

Figure 2: Night time

Tina dedicates 200 words to describing the importance that the night sky holds in her life. Tina describes the aesthetic pleasure of watching the stars, the contrast of the peace of the night compared with the pressure of the day, the space to think and the comfort that there is a visible connection to the people that she loves in the heavens. Tina, then, places a high value on the night sky but, as Tina suggests, it was not a subject that I would have broached without the direction of the map.

Tina: You probably would have mentioned the college and the driving… and my Mum's house obviously but you wouldn't have known anything about the way I feel about the night

Thus, even though I am an indigenous researcher Tina offers me an insight into aspects of her world that I would not have considered salient and reveals a subjective relationship with the night sky that I have no prior knowledge of.

Tina also describes how drawing her daughters acted to clarify aspects of the maternal relationship. When I looked at the picture I assumed that Chantelle had been presented as bigger than Louise simply because she is older and taller. However, Tina made clear that this was not the case and I could only understand the intended meaning of the image, with Tina's interpretation.

Figure 3: Chantelle and Louise

Tina: Louise she's small and if you look at that she ain't even in line with Chantelle because she's in the background and in my drawing I did that because sometimes I feel like Chantelle takes all the attention away from Louise

In order to gain an understanding of the internal narrative of the image, then, it is was imperative to acknowledge the role of the image-maker. My own interpretation of the visual data would have been inadequate for, both literally and metaphorically it is human beings who speak to one another and the lone image is an inadequate tool for understanding other people's worlds (Banks 2001). The practice of creating visual data, then, presented an opportunity for Tina to transcend the visible and actual physical difference by distorting generalities of alignment. My singular interpretation was still veiled by a web of taken for granted meanings but the combination of Tina's creativity and explanation contributed to a more nuanced understanding for both the researcher and the researched.

Ambiguity of familiarity

The danger that the study could be overshadowed by the enclosed, self-contained world of common understanding was not just restricted to the interpretation of the researcher. Chantelle, the youngest participant, illustrates this point with a picture of her home.

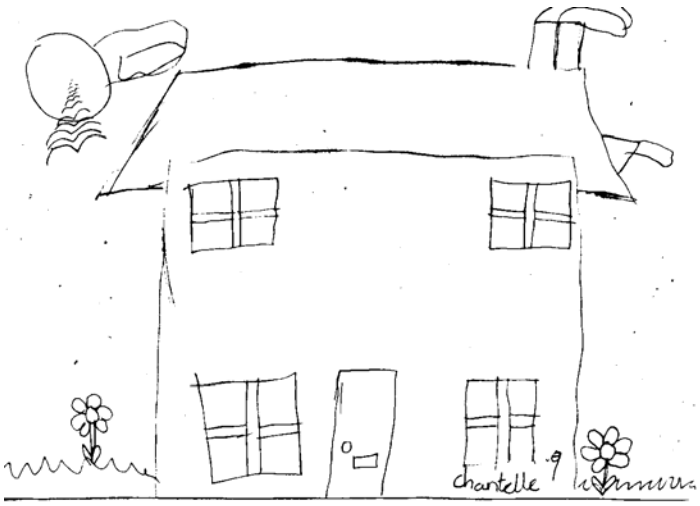

Figure 4: My House

Chantelle provided a visual representation of the outside of her house. The picture is interesting because it is the archetypal representation of a house common to childrens' drawings but it is not an accurate image of Chantelle's home, which is terraced with two windows at the front, no smoke coming out of the chimney and no flowers or trees in the front garden. Similar to the majority of visual tools, Chantelle's drawing is not an unambiguous record of reality (Ball and Smith 1992). When I take up the issue of the presence of the flowers in the picture with Chantelle she defends their inclusion.

I: So have you got flowers like this by your house? Did you just put them on or are they there?
Chantelle: Um we've got trees there but there are flowers outside my house but they're just starting, just starting to grow

The picture of the house then is a popular version of a house rather than a drawing that could be recognised as Chantelle's house. In this case then I am able to recognise discrepancies, nevertheless, the drawing illustrates that participants visual images may not always be accurate, which must be kept in mind when utilising visual data. When I have transcribed our conversation I speak with Chantelle again about these discrepancies. The structural incongruity is explained as necessary as there are two windows at the rear of the house, which Chantelle has moved to the front in her picture to take account of these rooms. Painting and sketching, then, as Damon (2000) maintains, are always dependent on the artistic ability of the author.

However, there is no attempt to elucidate the presence of the flowers in the garden. The inclusion of the flowers could be explained as an illustration of the gendered nature of children's drawing where young girls aspire to make their drawings pretty. Chantelle's drawing of her local environment was edged with gold stars and such elaboration is normative within the school setting. A correspondence between the drawing and school based activities is suggested with the use of the term *'work'* in Chantelle's description of generating the pictorial data.

Chantelle: I was happy when I did it (.) when I finished I was really happy because I was happy with my work so happy

Alternatively, the archetypal representation of home could be linked with Chantelle's desire to move house.

Interviewer: And what about your street what do you think about your street about the other houses and the road do you like it on your street?
Chantelle: No I hate it

Chantelle's aspiration to relocate is accompanied by a description of the preferred alternatives, which include larger houses with front gardens exhibiting flowers and shrubbery. This could suggest that Chantelle's subjective representation of her home provides an insight to the intersection of the social and the psychic, a visual representation of fantasy, hope and longing. Perhaps, this is an analytical leap but in either case the drawing makes departures from actuality that Chantelle feels she does not need to justify or explain.

This departure is not problematic in itself for the research was interested in Chantelle's own subjective account of the lived specificities of classed location at a particular time and in a particular place. However, Chantelle's account becomes problematic when incredulous at my continued interest she informs me that it does not matter because I have been to her house and I know what it looks like. This is resonant of the two-way taken-for-granted cultural competence that I spoke of earlier in the paper, a facet of my earlier research, which I hoped to suspend with a participatory methodology.

Akin to the participant in my undergraduate research, Chantelle communicated an assumption that I already understood her experience. Thus, the relative disadvantage inherent to my insider status remains and Chantelle may have drawn her maps with the proviso that I already 'know' the landscape of her subjective world. This illustrates an underlying and potentially deceptive assumption of shared understanding, which persists despite the adoption of self-directed visual methods.

Artistic Ability

When I asked Chantelle about what she thought about producing the drawings the confident reply was;

Chantelle: I found it pips thank you … I loved writing very neat writing

Chantelle's positive response to the task could be related to the similarity with school based activities where drawing is seen as an appropriate method for younger children (Barker and Weller 2003). Similar to other children who have engaged with visual data collection (Cook and Hess 2007), Chantelle also found that her illustrations acted as a support for her to tell her story;

Chantelle: Well really it refreshes you about what you really done and like you can say things and you can describe them because you can look at them and you can just say 'ah that's what I wanted to say now I remember'

In contrast to Chantelle, Tina, like adults in previous studies, lacked confidence in her artistic abilities and found the request to draw unconventional and alien to her everyday life (Kearney and Hyle 2004). Tina actually delayed the interview because she did not want to draw the maps or show me the drawings;

Tina: Yeah (.) it was just um (.) before I drew um I was just worried about what to do and what you'd think of them and that you know
Interviewer: Mmm
Tina: Because I'm so crap
Interviewer: 'Cause it's not normally what you do drawing
Tina: No it's not not drawing if you'd have asked me to do a five hundred word essay I could have done it in half an hour or something (laughs) (both laugh)

The lack of confidence in her drawing also led to omissions, one of these came to light in the interview but there may have been other items that Tina did not feel able to represent visually. In response to my question about her pets she replied;

Tina: Ah no I was going to draw them I can't draw (laughs) (both laugh) I should have put them in the corner

However, although Tina was reluctant to draw anything when I first mentioned the project when she actually completed the map making Tina was positive about the experience stating;

Tina: Drawings not really my favourite things to do but I have I've got to be honest I enjoyed doing this.

Conclusion

The adoption of self-directed mapping allowed time for Tina and Chantelle to reflect on their lives without the direction of an intrusive research voice. As a result both Tina and Chantelle created new connections and opened up experiences teaching me about places, spaces and identities, which I thought I knew. Visual methods of data collection, then, have been useful in making the familiar strange and interesting even though in many ways my own experience mirrors that of my participants. Furthermore, the mapping technique proved successful in engaging Chantelle and supporting her to tell her story. Despite her initial reluctance, Tina also reported enjoying the research experience. However, the adoption of visual methods can not be seen as a panacea for their remains scope for misreading, misinterpretation and misconstruction. Employing auteur theory proved essential in gain an understanding of the internal narrative of the image but there was still scope for omission and ambiguity.

The inherent value of self-directed visual data collation is that images, thus ideas are created without the intrusive presence of the researcher, which can be useful when the researcher is an insider who aspires to make the familiar strange. The use of mapping and the subsequent discussions tended to reveal far more than I would have expected using an entirely verbal approach for data collection. Employing these methods gave me a new insight into my participants' worlds as their maps led me to destinations beyond the confines of my preconceived understandings. Thus, the study made the familiar strange and interesting again.

Notes

1. The names Tina, Chantelle and Louise are fictitious and were chosen by the participants to maintain their anonymity.

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References

Ball, M. S. and Smith, G. W. H. 1992. Analyzing visual data. London: Sage.

Banks, M. 2001. Visual methods in social research. London: Sage.

Barker, J. and Weller, S. 2003. Is it fun? Developing child centered research methods. The International Journal of Sociology and Social Policy, 23 (1-2), pp. 33-58.

Belin, R. 2005. Photo-elicitation and the agricultural landscape: 'seeing' and 'telling' about farming, community and place. Visual Studies, 20 (1), pp. 56-68.

Cook, T. and Hess, E. 2007. What the camera sees and from whose perspective: fun methodologies for engaging children in enlightening adults. Childhood, 14, pp.29-45.

Damon, F. H. 2000. "To restore the events?" On the ethnography of Malinowski's photography. Visual Anthropology Review, 16 (1). Pp. 71-77.

Darbyshire, P., MacDougall, C. and Schiller, W. 2005. Multiple methods in qualitative research with children: more insight or just more? Qualitative Research, 5 (4), pp. 417-436.

Dodman, D. R. 2003. Shooting in the city: an autobiographical exploration of the urban environment in Kingston, Jamaica. Area, 35 (3), pp. 293-304.

Erikson, F. 1986. Qualitative methods in research on teaching. In Wittrock, M. C. (Ed.) Handbook of research on teaching. New York, NY: Macmillan.

Haylett, C. (2003) Culture, class and urban policy: reconsidering equality, Antipode, 35, No 1: pp 33-55.

Kaomea, J. 2003. Reading erasures and making the familiar strange: defamiliarising methods for research in formerly colonized and historically oppressed communities. Educational Researcher, 32 (2), pp. 14-25.

Kearney, K. S. and Hyle, A. E. 2004. Drawing out emotions: the use of participant-produced drawings in qualitative inquiry. Qualitative Research, 4 (3), pp. 361-382.

Lucey, H., Melody, J. and Walkerdine, V. 2003. Uneasy hybrids: psychological aspects of becoming educationally successful for working-class young women. Gender and Education, 15 (3), pp. 285-299.

Mitchell, L. M. 2006. Child centered? Thinking critically about children's drawings as a visual research method. Visual Anthropology Review, 22 (1), pp. 60-73.

Morrow, V. 2001. Using qualitative methods to elicit young people's perspectives on their environments: some ideas for community health initiatives. Health Education Research, 16 (3), pp. 255-268.

Pole, C. 2007. Researching children and fashion: an embodied ethnography. Childhood, 14, pp. 67-84.

Radnofsky, M. L. 1996. Qualitative models: visually representing complex data in an image/text balance. Qualitative Inquiry, 2 (4), pp. 385-410.

Rawlins, E. 2006. Mother knows best? Intergenerational notions of fashion and identity. Children's Geographies, 4 (3), pp. 359-377.

Reay, D. and Lucey, H. 2000. 'I don't really like it here but I don't want to be anywhere else': children and inner city council estates. Antipode, 32 (4), pp. 410-428.

Reay, D. 2004. 'Mostly roughs and toughs': social class, race and representation in inner city schooling, Sociology, 38, No 5: pp 1005-1023.

Rose, G. 2001. Visual methodologies. London: Sage.

Ross, N. J. 2005. Children's space. International Research in Geographical and Environmental Education, 14 (4), pp. 336-341.

Ross, N. J. 2002. Making space: children's social and environmental geographies. Unpublished Ph.D. thesis, University of Dundee.

Skeggs, B. 1997. Formations of class and gender. London: Sage.

Vrasidas, C. 2001. Interpretivism and symbolic interactionism: 'making the familiar strange and interesting again' in educational technology research. In Heinecke, W. and Willis, J. (Eds.) Research methods in educational technology, pp. 81-99.

Walkerdine, V., Lucey, H. and Melody, J. 2001. Growing up girl. Basingstoke: Palgrave.

Williamson, Howard 2004. The Milltown Boys Revisited. Oxford: Berg.

Exploring the relation between migration management and EU strategies for employment. An ethnographic study of the policy process at the European Parliament

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Abstract

The following article summarises the main results of an ethnographic study realized for my MSc dissertation on the European Union's migration and labour policies. The data was mainly collected through participant observation during a four months internship at the European Parliament. Starting from debate on globalisation, international migration and labour market governance, the major attempt of this work was to provide an ethnographic account of the "transnationalisation" of the EU governance of migration. The focus was on the labour dimension of the EU migration policy, exploring the relationship between this area of policy and the EU agenda for employment. While highlighting a changing articulation of competences between the EU, the Member States and new "transnational" actors in the management of migration, the findings suggest the effective, yet contradictory, emergence of transnationalisation processes at the level of the EU governance as well as in the area of migration policy. This piece deals specifically with employment issues. The major characteristic of the emerging transnational governance of migration lies in its selective nature, which attempts to build a regime of "conditioned mobility" for the movement of immigrants into Europe.

1. The theoretical debate on EU governance and transnational migration: an interdisciplinary trip through social geography, European studies and cultural anthropology.

The issue of transnational migration and its governance have been considered in different disciplines, from European studies to broader research in human geography, cultural anthropology and the sociological literature on globalisation (see Anthias, F., & Lazaridis, G. 2000, Burawoy 1991, Mitchell 1997, Ong 1999). More specifically European studies as well as mainstream approaches on international migration, consider the process of the "Europeanisation" of migration policy an emblematic case to explore the changing patterns in the management of international migration. These changes also implied a supposed decline of state territorial borders and national sovereignty (Cornelius W.- Hollifield J.-Martin P., 2004, Lahav 2004; Guiradon-Lahav 2000,. Sassen 1998).

Disciplines such as human geography and urban studies offer another important perspective to consider more broadly the issue of governance in the EU context, as well as the relationship between the employment aspects of the management of migration into Europe and the economic and social agenda of the EU. The "geography of spatial scale" in particular (Brenner 1998; Lefebrvre 1991; Harvey 1982; Castells 1972) provides crucial instruments to analyse the transformations of a specific form of governance such as the management of labour migration. This approach highlights the spatial "re-shaping" of European governance and the regulatory functions of the new policies. Indeed so-called regulation theory has characterised the theoretical debate in certain areas of human geography from the 1980s (see Peck and Tikell1992). According to the research on the new "geographies of labour market governance" (Peck 1994; 1996) the relationship between areas of social and employment policy assume a particular form among neo-liberal states, which are increasingly responding to globalisation by "hacking at the roots of welfare state while framing economic policy solely in supply side terms" (Peck 1996, p.185).

Importantly, for my research these studies emphasised an intrinsic ambivalence in both the spatial and the regulatory dimensions of the EU governance. On the one hand state-rescaling can in fact be viewed as a strategy of de-regulation undermining the welfare functions of local governments (hence neglecting the typical functions of the "Fordist –Keynesian era). On the other hand a strategy of "re-regulation" seems to emerge in terms of new "institutional capacities" aimed at promoting world-competitiveness between urban centres and developing new forms of welfare such as "workfare policies" and public-private partnerships (Cerny 1995,

cit. in Brenner 1999 p.440). In particular, focusing on the role of the state in the Post-Fordist era of capital accumulation, Jessop (1994) highlights the "shift toward internationalised, flexible (but also regionalised) productions system" producing a de-facto weakening of the state powers and loss of authority, and at the same time "both the need for supranational coordination and the space for subnational resurgence" (p.264).
Therefore, besides the issue of "re-scaling" state territoriality, social geographers crucially point to the "regulatory" reasons at the base of the emerging institutional configurations. In this way they reveal a further dimension of the processes of transnationalisation that sits at the core of my inquiry.

Within the major European studies of migration policy, some describe the changes in the political and economic functions of the state as processes of transnationalisation, interpreting them as "counter strategies" against the restrictive pattern of migration policy arising from the process of Europeanisation (Lahav 2004). Following a different approach, my study draws on the perspective of the "autonomy of migration" (Mezzadra 2006) and the critical thesis recently developed on transnational migration and the "European migration regime" (Karakayali – Tsianos 2005; Papadopoulos, Stephenson, Tsianos 2008, Rigo 2005). Along with new approaches on transnational migration coming from cultural anthropology (Ong 1999) these perspectives provide interesting element to consider the European regime as emerging from the everyday practices of immigrants.

These critical approaches position themselves at a distance from the mainstream "migration studies" originally developed in the US but increasingly spread among European scholars. The sociological literature on migration in Europe, while maintaining the nation state at its core, primarily considers the effects of migration on the national demographic and social composition. Therefore it focuses on the efforts that states should endorse for the "management" of the "flows" of refugees and immigrant workers (for a critical review see Ong 1999, p.8). This approach interprets migration under a "structuralist" perspective as the result of "objective" push and pull factors, explaining the movement of migrants either in terms of flows induced by the gaps between "core" and "periphery" countries ("world-system" theory) or merely in terms of "labour supply…flowing toward an advanced capitalist formation" (following the neoclassical economic theory) (Mezzadra 2006, p.185).

On the contrary the "autonomy of migration perspective", without escaping an analysis of the labour market, suggests the need to move beyond an interpretation of migration focused on the state level. Rather, the autonomy of migration looks into the crisis of the national labour market from the point of view of the current transformations addressing its "national order". The crisis mostly concerns the production of the labour supply within the traditional territorial boundaries of the nation state, thus configuring the search for a new mix of labour mobility and immobility as the main concern of contemporary migration policies, at both national and supranational level (Mezzadra 2006, Duevell 2002).

The very change of terminology from "immigrant" to "migrant" in the literature signals in fact a more profound change in the understanding of international migration in sociology and anthropology. Since the early 1990's the concept of "immigrant" has been called into question by American anthropologists as unable to explain the "simultaneous embeddedness" that current immigrants construct in both their societies of origin and destination (Glick Schiller, Bash and Blanc 1999). Various aspects related to the restructuring of capital globally, as well as a relatively decreased significance of national boundaries in the distribution of resources and productions, contributed to give a new shape to current processes of migration. These cannot be described anymore as "one-time" and "unidirectional" (Pries 1999, p.26), rather they must be seen as constituting a new social group in a new "social field", which goes beyond its geographical extension. The "transnational social spaces" described by the new literature encompass the multi-stranded social relations that connect migrants' lives across borders. As a result, the qualitative change in current forms of migration points to a parallel shift of attention from the integration of immigrants in the society of arrival, to the acknowledgment of the "multiple, multidirectional, incremental territorial migrations related to employment or specific life phases" (p.27). Re-thinking the concept of "immigrant" within a new anthropology of transnational migration was therefore intended to unveil the political factors that have shaped the image of immigrants as "uprooted" subjects, supposedly ready to be assimilated and become loyal to the new community of residence.

More recently American sociological studies such as the "new economies of migration" (Massey et al. 1993) draws from the idea of transnational migration, and has emphasised the impact of familiar and communitarian networks upon the different phases of the migration processes. In this way they started to recognize some forms of "agency" on the part of the immigrants. However, even the new approaches ultimately risk re-proposing a general theory of "social

integration" (Mezzadra 2006, p.186). On the contrary the "autonomy of migration", brings back into the framework of analysis not only the real processes of exclusion and stigmatisation produced by the new systems of governance of mobility, but also the struggles and everyday practices of the immigrants as important factors shaping these systems. Only by questioning assumptions of the social and cultural integration of settled or unsettled immigrants, is it possible to analyse the contemporary migration processes from the point of view of the subjective practices of the migrants. Indeed these practices "exceed" the "objectives factors" of supply and demand supposedly at the base of migrants' mobility as understood in structuralist approaches. This means migration is interpreted as a social fact that transcends the mere functioning of the labour market (p. 185).

At the same time the "autonomy of migration" looks also at the forms of "selective inclusion" arising from a new system of governance that has acquired the form of a proper "European migration regime" (Karakayali –Tsianos 2005; Papadopoulos-Stephenson,-Tsianos, 2008, Rigo 2005). This involves complex patterns of "externalisation" and "internalisation" of control technologies, determining a series of profound implications for the "institution of the border" itself and EU membership (Rigo 2005). The so called transitional measures applied to the citizens of the Eastern European "accession countries" limited their rights to freely circulate and to search for employment in the rest of the EU (Rigo 2005, p.13). In this sense, these ad hoc measures represent an emblematic case of the overall process of differentiation produced by the EU migration regime, which mainly works by configuring a "plurality of diachronically differentiated legal positions" (p.11) in the form of the EU membership.

However, the point of view of the autonomy of migration interprets such an ambiguous movement of "de-territorialisation" and "relocalisation" of the European borders management (p.14) as stemming from the everyday "practices of citizenships" expressed by immigrants. The potential inclusiveness of a system that paradoxically follows their mobility in the attempt to limit such movements, is in fact embodied by the immigrants themselves, who concretely contest the EU "fortified borders". The emphasis on migrants' everyday strategies to cross the borders suggests a move away from "an abstract model of post-national citizenship" as something naturally liberating and inclusive (p.14).

The contradictory trends currently involving the European space therefore lead us to question the use of the term "post-national" to describe the new articulations of EU migration

management and the role of the agency of immigrants in shaping it. Moreover anthropological studies on "transnationality" introduce a further reflection on the effects of immigrants' practices and their interaction with the strategies of governance, bringing back to the debate the issue of labour mobility.

By highlighting the patterns of mobility and flexibility at the core of the contemporary "cultural logics of capital accumulation" (Ong 1999, p.6) the concept of transnationality developed by Aihwa Ong provides a further understanding of the re-articulation of global economy with the multiple political, ethnic, and personal identities acting at the borders. Yet, this process does not necessarily imply a loss of control from the part of the state over its subjects or a falling significance of its political borders' (Ong 1999, p.3). In fact, while most of the critical research on the European migration policy identifies a tendency of "de-territorialisation" of state sovereignty as an effect of the process of communitarization (Lahav-Guiradon 2000, Rigo 2005), other authors provide a different interpretation. Papadopoulos, Stephenson and Tsianos (2008), explore the transformations of EU policy for the control of migrants mobility in order to warn that such a "postnational process of border displacement" should not be interpreted as a mere extension of the sovereign power of the state, which simply delegates to external agents the control over its borders: "rather it represents a multifaceted, constitutive plane of struggle, where the regime of mobility control is itself challenged by the new fluid, streamlined, clandestine, multidirectional, multipositional and context-dependent forms of mobility" (p157). Consequently, the choice of the term "regime" instead of "system" emphasises the relation between migrants and the new "agents of control" in the determination of the regime itself. The main feature of what the authors call the "Liminal Institution of Porocracy" of the European migration regime lies in the "flexibility" or "porosity" of the management of labour migration, which mirrors precisely the flexibility of immigrants and their capacities of adaptation within the context of border crossing (p.160).

Nevertheless, governance and regulation remain crucial aspects in the process of Europeanisation of migration policy. The EU migration regime is still inserted in a regulatory framework in which sovereignty is de-territorialised but continues to use the borders to produce differentiated spaces of mobility and legal standards. This is exemplified by the 'Schengen' space itself, which remains dominated by a "governance of differential homogeneity" (p.171). In this sense post-national instances are continuously contradicted by "the real politik of regulation" (ibid). The failure of the attempts to extend the process of communitarisation to other areas of

migration policy (such as employment and residence rights for long term immigrants) under EU competences may also mirror these two contradictory trends.

The focus of my research: the labour dimension of EU migration management and its transantional configuration

As this overview has shown, while only few research studies specifically address the links between migration policy and the broader government of labour mobility, even less explore the relation between labour and migration policies. The critical literature underlines the interaction between the new strategies for the management of mobility and the everyday practices of migrants as a crucial point of view from which to observe the broader transformations at the level of European governance. While sharing these substantial assumptions, my study looked more specifically at the relationship between labour and migration as it is intended and presented within the EU process of policy making. The major aim of my research was is in fact to identify the different ways in which the transnationalisation of sovereign power and economic governance is declared and accomplished through the migration policies of the EU.

In particular, my research questions addressed three dimensions of the relationship between labour and migration. Firstly, drawing from the debate on the European migration regime, I investigated to what extent the process of "externalisation" of migration policies assumes a transnational configuration with explicit regard to issues of employment.

Secondly, I explored the impact of the proposed migration policies on the internal power articulation of EU governance, hence addressing the relationship between Union and Member States. I investigated the tensions between processes of communitarization and the resistance of nation state to relinquish their prerogatives on migration matters. In other words I asked: is the rejection of the "zero-sum game" (Weiler 2003) in the new geometry of migration governance emblematically expressed by the current European policy, and if so, in which forms?

A third dimension concerned more specifically with patterns of employment within the EU migration policies and their relationships to the current European strategies for employment and economic growth, called the "Lisbon Strategy". This agenda in fact includes a range of issues, such as the "supply side policies", which are considered in relation to the demographic impact of increased migration into Europe. Discourses of labour market flexibility, the labour market

integration of migrant workers and finally the policies linking the fight against undeclared work with clandestine migration also represent crucial connections between the two policy areas.

2. The methodology: an ethnographic approach to Grounded Theory

This research is an ethnographic study on the EU policy making carried out during a four month internship at the European Parliament. The data of this research have been mainly collected through participant observation in the European Parliament in Brussels. My position as an intern at the Italian delegation of the PSE offered me the opportunity to follow the every day debate and policy making on EU migration policies. It gave me access to the most up to date documentary material on the issues of labour and migration management, from both the Parliament and the European Commission.

In general terms the research design draws upon the perspective of participant observation elaborated by Michael Burawoy (1991; 1998; 2000), who emphasises the possibility for ethnographic research to illuminate "the macro world through the way the latter shape and in turn is shaped and conditioned by the micro world, the everyday world of face to face interaction" (Burawoy 1991, p. 6). Challenging the traditional limits of ethnography as restricted to "micro and a-historical sociology", the "unbounded ethnography" proposed by Burawoy (1991) moves towards the "reconstruction of theory" on the base of what the researcher has learned in the "dialogue" with the participants instead of focusing on detailed and descriptive accounts of people's speech and actions. This approach fits particularly well with my study, which draws from an experience of participant observation among the EU institutions to explore a more theoretical question regarding processes of transnationalisation in the governance of labour and migration.

I considered the critiques proposed by Burawoy (p.1991, p.275) which regard the tendency of grounded theory towards generalisation, which risks overlooking "the specificity" of each situation. However, in the collection and analysis of data I followed the general indications of grounded theory (Glaser and Strauss1967) while drawing more specifically on the "ethnographic approach to grounded theory" developed by Charmaz and Mitchell (2001). I found that adopting both ethnographic fieldwork and grounded theory as complementary methods was particularly appropriate for my research. In fact it entailed participant observation (see the definition of Hustler 2005, p.16) within the European Parliament while utilising (as main sources of data) policy documents and interviews with key informants collected during the fieldwork. The

perspective of Charmaz and Mitchell allowed me to combine the pattern of "theory development" typical of grounded theory with the sensitiveness of ethnographic research (Charmaz-Mitchell 2001, p.160) (i.e. the European Parliament in my case).

Therefore the research primarily followed the fundamental indications provided by Glaser and Strauss (1967): simultaneous involvement in data collection and analysis, development of codes and categories directly from the data (and not from pre-given hypothesis), theoretical sampling aimed at theory construction (rather than for population representativeness) and continuous development of theory at each phase of data collection and analysis (Glaser and Strauss 1967, cit. in Charmaz-Mitchell 2001, p.161). However, the categories, concepts and theoretical level of analysis have been mainly understood as emerging "from the researchers' interaction within the field and questions about the data" (Charmaz 2000: 522). In particular ethnographic research and grounded theory have worked as useful complementary methods in order to explore both the policy discourse of the EU on labour and migration in the documents and to elaborate on the data analysis, while observing and participating in the every-day policy process of the Parliament.

With regard to the qualitative data analysis, Grounded Theory (Glaser and Strauss 1967) has been followed as the broad method for the analysis of both the documents and the interviews' transcripts. Moreover, the very choice of the ethnographic study induced me to adopt the ethnographic approach to coding in Grounded Theory elaborated by Charmaz and Mitchell (2001). However, the specific nature of EU policy documents on migration induced me to employ additional methods, namely narrative analysis (Coffey and Atkinsons 1996) for the policy papers, and discourse analysis (Potter and Wetherell 1994; 1995) for the interviews. Here, my major interest consisted in identifying the "patterns" emerging from both the documents and the interviews in order to develop "middle range theory" (Charmaz and Mitchell 2001, p. 165). Nonetheless, a focus on the "functions" (Atkinson and Coffey 1996, p.62) of the narratives added an important insight in identifying the main devices used by both the documents and the respondents to justify and "accomplish" a certain policy measure rather than another.

3. Transnationalising the EU governance of labour and migration

The ethnographic study of the European Parliament revealed a substantial separation between the areas of migration and labour policies. The distinction was mainly exemplified by the lack of

cooperation between the two committees dealing with these policies within the Parliament (namely between the Committee on Civil Liberties, Justice and Internal Affairs and the Committee on Employment and Social Affairs). On the one hand the missing cooperation at the level of everyday EU policy-making might be interpreted as the sign of continued resistance on the part of member states. They in fact understand migration primarily as a matter of national security and internal affairs. On the other hand, the increased importance of the employment dimension within the EU migration policy emerged both from the analyses of policy documents and from the interviews with the EU officers and MPs.

These contradictory findings have important implications for understanding the relationship between employment and migration policies as well as for the new forms of governance emerging at the EU level. The results of the ethnographic study have been therefore discussed in relation to the theories on transnationalisation, social regulation and labour market governance, which emerged from the literature on globalisation in general and on the Europeanisation of migration policy in particular.

The external dimension of migration management: a new system of "conditioned mobility"

The analysis of the new EU policies regulating the admission channels for non-EU migrant workers and the management of migration flows between the EU and "third countries" revealed the fundamental relevance of the employment dimension within the EU approach on migration policy. First of all, the Policy Plan of the European Commission (CEC 2005) gives priority to the so called "legal migration agenda" as a means both to reduce illegal flows and to respond to the labour needs of the European labour markets. Nonetheless, what I described as the "category-based approach" emerging throughout the different directives, points to a selective system for the management of labour migration into the EU. In fact, both the comparative analysis of the two crucial documents on admission matters, (i.e. the Policy Plan and the communication on "circular migration" and "mobility partnerships") and the interviews confirmed a trend that generally favours the mobility of immigrants. Yet, the intention seems to facilitate mobility but according to temporary patterns and other pre-conditions dictated by the security and labour supply concerns of the member states. More specifically, the proposed "circular migration schemes" introduce further channels for the selection of immigrants attempting to "normalise" the circularity of highly mobile workers coming from outside the EU. In fact, beside the overall

condition of temporary settlement, privileged channels are created both according to the occupational category of immigrants (e.g. from "highly skilled" to "remunerated trainees") and on the basis of the country of origin. In sum, circular migration and mobility partnerships should be promoted under certain conditions, primarily with those third countries that cooperate in the fight against "illegal immigration".

Such a system of selection for the admission of migrant workers is well synthesised by the pattern of differentiation that Rigo (2005) indicates as one of the main feature of the "Europeanisation" of migration policy. The circular migration schemes aim in fact at governing the movement of immigrants back and forth to the EU, treating them differently in accordance to their legal position, e.g. whether they are already settled immigrants or seasonal workers. Moreover, apart from the juridical status, further channels of preference emerged concerning the employment and the occupational position of non-EU immigrants.

However, it is the analysis of the "external dimension" of EU migration regime (as it emerges from the proposal on circular migration and mobility partnerships) that which signals the emergence of a specific employment dimension within the process of transnationalisation of migration policies. In fact, the emphasis on "incentives" and on the "policy of return" as core elements of the mobility partnerships with third countries confirm a general interest in facilitating the mobility of immigrant workers, yet according to selective and differential patterns. In fact, the "Return policy" constitutes a central condition at the base of circular migration and mobility partnerships, despite their emphasis on development and labour issues. Indeed, the blurring differences between the internal and external dimensions of migration policy, foreign and internal affairs are clearly exemplified by the wider concept of "border management" supported by the Commission officers. As emerged in particular in the interview with them, the interest in managing migration in partnership with third countries includes both the need to "export" labour (when it is no more needed) and to cooperate in the fight against illegal migration. Furthermore, the creation of new transnational job agencies in the countries of origin also brings evidence of the increased relevance of labour issues in the relation to the "external dimension" of migration management. However, the accent on return migration and the readmission of illegal migrants as basic conditions for third countries to join mobility partnerships might reveal a fundamental interest of the EU in creating a system that favours the partial or "conditioned" mobility of third country nationals.

In this sense, the findings confirm the thesis of the "autonomy of migration" perspective, which highlights the emergent "new mix of labour mobility and immobility" at the core of the new "European regime" for the governance of migration (Karakayali –Tsianos 2005; Papadopoulos, Stephenson, Tsianos 2008, Mezzadra 2006). In fact, the strategies of "selective inclusion" at the base of the processes of externalisation and internalisation of the European migration regime and the characteristics of "porosity" and "flexibility" of the EU borders indicated by these authors clearly appeared in the new forms of migration management highlighted in this research. However, the analysis revealed new features in the governance of migrants' mobility, pointing to processes of transnationalisation regarding specifically the area of migration policy related to labour issues. Some other concepts offered by regulation theory and social geography represented further tools to interpret the regime of selectivity that emerged in my research, providing particular insights into the relationship between migration policy and the European strategy for growth and employment.

Regulating migrant labour between concerns of labour supply and social cohesion

Theories of labour market regulation and the geography of spatial scale can be particularly useful in exploring the role that migration plays within the EU strategy for employment (the "Lisbon agenda") and its revised policy Guidelines. In fact the geographic approaches to economic and labour market governance under the crisis of the welfare state in Europe, stress different aspects of current forms of labour regulation. Although not explicitly focussed on migration, but generally on matters of labour market governance, they studied the transfer of state competences and powers upward to supranational actors and institutions (Brenner 1998, 1999; Jessop 1994, 2003).

My research addressed the new strategies of governance of labour migration, considering them in relation to the broader patterns of employment regulation of the "Lisbon agenda". More specifically, the Employment Guidelines appear to recognise explicitly the important contribution of migration to Europe, in relation to three fundamental objectives: increasing labour supply and retaining more people in employment, combating demographic deficit while improving the "matching of labour market needs" (Guideline N 20 EC 2005) (along with the interest in fostering flexibility and competitiveness overall in the European labour markets), and, finally, the fight against undeclared work as a strategy to combat labour market segmentation (EC 2005). Hence the policies emerging from the Lisbon strategies are intended to increase the

provision of labour also through the labour market integration of "legally staying" immigrants. More generally the Guidelines emphasise the positive impact of a well-managed migration into Europe, according to a policy able to counteract demographic trends such as the contemporary ageing of the EU workforce.

The centrality reserved for concerns regarding 'supply' (by both employment and migration policy documents) uphold the interpretation offered by regulation theories about the European process of integration. In fact, as Jessop argued (1994, p.275) the EU economic and employment governance seems to be mainly oriented toward "supply side" measures aimed at fostering structural competitiveness and individual employability (see also Aschiagbor 2005). However, beyond the concern about the control of labour supply and its reproduction (Peck 1994) the social regulation approach interestingly highlights the concept of "crisis management" (Jessop 1994, p.275) at the origin of the state's regulatory intervention and its crucial role as mediator of the tensions emerging from global markets. Underlying assumptions are that crisis management and instability are continuous and intrinsic characteristics of the very forms of labour market and social regulation (Peck and Tickell 1994). In this sense the new forms of regulation proposed by the EU strategy for growth and employment might indicate a repositioning of this central political function traditionally held by the state, upward to the supranational level. by following a complex process of displacement of regulatory competences throughout the different levels of governance (being they local, regional, national or supranational) (Brenner 1999).

The concepts of instability and crisis management illuminate particularly the second policy priority that EU migration shares with the employment policy, that which proposes to use migration management appropriately in order to improve the matching of labour supply and demand. For instance, one of the employment objectives of circular migration and mobility partnerships is to increase the capacity of predicting labour shortages in cooperation with third countries (CEC 2007, p.8). Nonetheless, the analysis of the proposals revealed a major tension between the need to predict labour and skills shortages on the one side and the fluctuating demand for labour (a typical trait of labour in developed countries) on the other.

According to the perspective of the "autonomy of migration" one could argue that, rather than responding to the constitutive flexibility of labour demand in the EU countries, the flexibility of the new schemes is induced exactly by the continuous movement of transnational immigrants across the borders. In this sense the "turbulence" (Papastergiadis 2000) represented by the

mobility of immigrants could be interpreted as emblematic of the same instability and continuous crisis detected by regulationists at the core of the European strategies for employment and social regulation. The focus of this study on the internal EU policy discourse of labour and migration does not allow for elaboration on the different aspects and nature of the unpredictability and instability represented by transnational migration, e.g. the cultural logics at the base of transnationalisation process (Ong 1999). However, by underlining these elements of instability the present research places itself at a distance from the neoclassical reductionism of migration as merely following structural factors of supply and demand.

Finally, the interrelation made by the Guidelines between the fight against undeclared work, and the employment of "illegally staying immigrants" (CEC 2007a), seems to contradict the confinement of the undeclared work of immigrants to "illegal migration" matters. The comparative analysis of the documents and interviews indicated how this trend may reflect the belief of the Commission in the fundamental separation between immigration policy, on the one hand, and labour and social policy on the other (CEC 2007a, p.2). In fact, such a distinction could eventually been used to justify the punitive approach of the directive (prescribing penal measures against employers of "illegal immigrants") while assuring Member States that repressive measures toward immigrants are respectively prescribed by the "Return directive" (CEC 2007a, p.2). The persisting "securitarian approach" toward so called illegal migration may be considered strategic by a system of governance that continuously pursues the mobility of migrant workers while at the same time seeking to limit it.

From the management of migration to the question of state sovereignty: "re-scaling" the EU governance of migration between national, supranational and new transnational actors

A further specific objective of my research concerned the effects of the "Europeanisation" of migration policies in relation to the broader transformations regarding the EU system of governance. In particular, moving from the critical debate about a supposed weakening of state sovereignty as a consequence of the processes of communitarisation and de-territorialisation (Ong 1999, Rigo 2005) the question was whether the "zero-sum game" model was still able to explain the new configurations of sovereignty, as they emerge in the case of the EU governance of migration. Indeed, the analysis of the policy documents and the interviews with EU policy makers and MPs indicate a pattern of increasing regulation on the part of the supranational

bodies, which may be driven by a major interest in securing the provision of labour supply to the European markets. However, the "zero-sum model" seemed not able to describe the new forms of this regulation, which might appear to be more complex than a mechanic process in which switching state's competences upward to the supranational level implies an equal loss of state sovereignty. On the contrary the findings show how the state retains an important role in the governance of migration. A telling example is that the final decision about the actual number of immigrants admitted into the national territory remains a prerogative of Member States. Furthermore bilateral agreements between individual member states and countries of origin continue to occupy a central position within the new transnational architecture of the envisaged mobility partnerships (Bou, int.n1, FSJ, Commission).

Why then is it important to underline the transnational character of the new EU governance of labour migration? In fact, both the proposals of the Commission and the interviews revealed the emergence of new actors in the management of labour migration, identifying a "multilevel regulatory framework" cohabited by EU, national and bi-lateral forms of intervention. The interviews in particular indicated a fundamental matching between "traditional" bilateral agreements between Member States and third countries for the "exchange" of migrant labour across the EU borders, and the role of new "transnational agencies" such as the "cross-border labour intermediation services" as well as the "Migration Support Teams" which are to be introduced by the new "mobility partnerships" (CEC 2007, p.6).

More specifically the objectives pursued by the mobility partnerships and the overall system of circular migration contribute to a multilevel framework where:
Third countries could be encouraged to put in place legal and administrative arrangements to facilitate circular migration. Partner countries should in particular commit themselves to improving employment and labour matching services, as well as vocational and language training or the ability to foresee skill needs and supplies and provide information on labour needs abroad. Partner countries could also be supported in enabling returning migrants to contribute effectively to their home societies (CEC 2007, p. 13)

The two "pilot projects" currently managed by the Commission in Mali and Morocco are concrete examples of an initial "externalisation" of migration management to third countries. As revealed by the interview with a policy officer in the Commission's Director General RELEX (External Relation), the experiment in Mali in particular shows the multiple dimension of the EU management. The project includes various issues ranging from labour mobility to the fight

against illegal migration and readmission agreements. However, according to another policy officer of the Commission from the DG EuropeAid dealing with issues of development and cooperation, the intention of the new mobility partnerships is to initiate greater collaboration with the labour ministers of third countries. Ideally each country should have a system to manage labour flows in order to "prepare their citizens for the global labour market". (Genetzke int. n° 5, EuropeAid, Commission). Therefore the issue of mobility re-emerges at the core of the transnational architecture for the governance of migration, in accordance with the major objectives of the Lisbon Agenda for Growth and Employment. The major goal of the partnerships is to anticipate labour mismatches and provide the needed supply of labour, while confirming the centrality of the eventual return of immigrants to their countries of origin.

Apart from highlighting the emergence of transnational agencies specifically tackling issues of labour migration, the interviews introduced the notion that increased intervention and regulation at the EU level could eventually determine the strengthening of state capacity. For instance, by playing the new international job matching centres against the widespread activities of existing private agencies (which often facilitate informal and often illegal trafficking) (Genetzke, int. n° 5, Europaid, Commission). Therefore the proposal on migration and mobility partnerships may be finally intended to favour a new form of public regulation, whereas the supranational level enhances the capacity of member states to regulate already existing transnational private agencies. Here the intention may be that of maintaining a certain level of protection for migrants workers and a certain level of social cohesion overall in the states of the EU, while assuring all the aspects of "integrated borders management" work (including the fight against illegal migration). Whether or not the attempt to build up transnational agencies controlled by Member States and supervised by the EU confirms such an intention remains an open issue. This trend in fact cohabits with the opposite one toward new practices of externalisation and "privatisation" characterising the European governance of migration (see CEC 2007a).

Despite these contradictory trends, the new character of the EU governance of migration is clear: while the Member States through bilateral agreements still define the quotas of immigrants being accepted, the EU will ultimately exert its influence by deciding which countries will join the mobility partnerships and establishing the general criteria of entry (e. g. according to the so called "Neighbouring policy" with countries in Eastern Europe and northern Africa, "regional interests" furnish the base for initiating readmission agreements) (Bou, int.n° 1, FSJ, Commission). Beside the supranational level, the fact that new actors gradually enter the

framework for the management of labour mobility, through public and private agencies in the countries of origin, also highlights a pattern of transnationalisation in the EU governance of migration.

Therefore, as argued by Papadopoulos, Stephenson and Tsianos (2008), the emerging process of power displacement in the European migration regime cannot be reduced to a simple reinforcement or extension of state powers. Instead of a mere delegation of sovereign capacities (the concept of "regime" endorsed by these authors) crucially helps to identify the multiplicity of actors and level of governance emerged in the research,. This does rather highlight a "third space" of "negotiation" (p.165) to which different subjects and driving forces participate, often following conflicting logics.

Similarly, the concepts of the "re-scaling" of powers provided by Brenner (1999) to describe the new spaces of social regulation within human geography is useful to comprehend the transformation of EU governance emerging from the Europeanisation of migration policy. In contrast to a "statist" position, it is argues that the "national scale" is itself subject to a process of profound redefinition as a consequence of "new institutions and regulatory forms" arising at both supranational and local scale under the neo-liberal agenda (Brenner 1999, p. 439). Although with a different accent, the perspective of the autonomy of migration also highlights the persisting importance of regulatory concerns in the EU management of migration, which continuously confronts current processes of transnationalisation embodied both by the EU governmental strategies and the everyday practices of mobility of the migrants.

Conclusions

In summary, both the autonomy of migration and the regulation perspectives enlighten the main findings of my study on the European management of migration and its relationship with labour policy. The research provides some evidence regarding the emergence of a transnationalisation process (also for that specific point of intersection between labour and migration) as they are explicitly presented by the policy discourse of the EU. In particular, a pattern of "flexible regulation" might synthesise the new European governance, in that it mediates between national and supranational instances of regulation while reflecting the very flexibility and mobility embodied by transnational migrants.

The increased importance attributed to labour issues in relation to migration management would furthermore indicate the attempt to create a system for a transnational allocation of labour, especially by means of anticipating labour and skills gaps in cooperation with the countries of origin. Following this objective, the outlined regime for the governance of migration seems to be based on a fundamental contradiction: it maintains the traditional form of bilateral agreement of labour flows (able to provide "secure" circular and return migration) within a framework which has yet become transnational, i.e. regulated by a supranational framework where both states, as well as formal and informal transnational agencies actively participate, continuously searching for an original mix between mobility and fixity.

This research has provided a step forward in the understanding of the complex dynamics involved in the new articulation of the governance of labour and migration in the EU. However, if this study could be repeated, the attempt would be to develop a more grounded ethnographic account of the EU migration and labour policy, thus providing a deeper understanding of the very process of regulation and policy production. Furthermore, comparative research directly involving migrant workers and their experiences, would provide opportunities to investigate the interaction between the new strategies of the EU governance of migration and the everyday lives and practices of transnational immigrants. Instead of looking exclusively at the policy side of the phenomenon leaving aside the perspective of the migrants who are actually the subjects of these policies and regulation, other approaches can be provide useful insights. For instance the "situated ethnographies" conducted by some researchers on the "feminisation of labour regimes" across transnational spaces (see Ong 1999, p.11), would provide a further interesting perspective to consider at the same time as the transformation of strategies of governance in relation to macro political economic of cultural processes and individual practices. More broadly, feminist approaches to political economy could add a focus on the ways in which the cultural logics of transnational practices are embroiled within the more structural principles shaping the different forms and "geographies" of capitalism (Mitchell 1997), thus exploring more deeply the relationship between current process of transnationalisation, the new forms of social regulation emerging under globalisation and their "spatial" dimension.

Moreover the findings regarding the emerging forms of "flexible regulation" at the level of the EU governance and the tensions identified between private, informal and public agencies in the management of labour and migration, encourage future research on the contradictory relationship between "regulation" and "deregulation" strategies of governance (Aschiagbor,

2005). More generally the relation between simultaneous processes of increased regulation and "informalisation" in the global labour market should be further enquired (Sassen 1997). Indeed, these latter trends can be interpreted along with the increasing patterns of selectivity and differentiation of the EU "porous" borders, further exploring the new tensions characterising the employment dimension of migration policy as they emerged from my challenging experience of participation within the EU process of policy making.

Bibliography:

Anthias, F., & Lazaridis, G. (2000). *Gender and migration in southern Europe : women on the move.* Oxford: Berg.

Appadurai, A. (1996). *Modernity at large: Cultural dimensions of Globalisation.* Minneapolis: University of Minnesota Press.

Aschiagbor, D. (2005). *The European Employment Strategy. Labour Market Regulation and New Governance.* Oxford: Oxford University Press

Atkinson, P., and Coffey, A. (1997). Analysing Documentary Realities. In D. Silverman, (Ed.), *Qualitative Research: Theory, Method and Practice.* London: Sage.

Atkinson, P. - Coffey, A.- Delamont S. (2003). *Key Themes in Qualitative Research, continuities and changes.* Walnut Creek, Calif.: AltaMira Press

Bauböck, R., and Rundell, J. (Eds.) (1998). *Blurred Boundaries: Migration, Ethnicity, Citizenship.* Aldershot: Ashgate.

Beck, U. and Grande, E. (2004). *Das kosmopolitische Europa.* Frankfurt a. M.: Suhrkamp

Bell, C. (1969). A Note on Participant Observation, *Sociology*, 3: 417-18.

Beynon, H., D. and Grimshaw, et al., (Eds.). (2002). *Managing employment change. The new realities of work.* Oxford: Oxford University Press.

Bigo, D., and Guild, E. (2003). Le visa Schengen: expression d'une stratégie de «police» à distance, *Cultures & Conflits*. 49-50

Boltanski, L. and Chiapello, E. (1999). *Le nouvel esprit du capitalisme*. Paris: Gallimard.

Brenner, N. (1999). Globalisation as Re-terriotrialisation: the Re-scaling of Urban Governance in the European Union. *Urban Studies*, 36 (3): 431-451.

Brenner, N. (1998) Between fixity and motion: accumulation, territorial organization and the historical geography of spatial scales. *Environment and Planning D*: Society and Space, 16: 459-481.

Bryman, A. (2003). *Social Research Methods.* Oxford/New York: Oxford University Press

Buroway, M. (1998) The Extended Case Methods. *Sociological Theory* 16(1): 4-33.

Burawoy, M., A. and Burton, et al., (Eds.) (1991). *Ethnography Unbound, Power and Resistance in the Modern Metropolis*. Berkeley Los Angeles/London: University of California Press.

Castells, M. (1996). *The Rise of the Network Society.* Cambridge, MA: Blackwell.

Castells, M. (1972). *The Urban Question*. Cambridge, MA: MIT Press.

Castles, S. and Miller, M. J. (2003). *The Age of Migration. International Population Movements in the Modern World* (Third Edition). New York – London: The Guilford Press.

Castles S., (2004). Why Migration policies fail. *Ethnic and racial Studies*, Vol. 27 No.2 March 2004 205-227

Charmaz, K. (2000). Grounded Theory: Objectivist and Constructivist Methods. In N.K. Denzin, and Y.S Lincoln, (Eds.), *Handbook of Interview Research: Context and Method.* California: Sage.

Charmaz, K. (2004). Grounded Theory. In M.S. Lewi-Beck, A. Bryman and T. F. Liao (Eds.), *The Sage Encyclopedia of Social Science Research Methods* (Vol 1-3) Thousand Oaks, Calif. : Sage

Charmaz, K and Mitchell G. (2001). Grounded Theory in Ethnography. In P. Atkinson, et al. (Eds.), *Handbook of Ethnography*. London: Sage.

Coffey, A and Atkinson, P. (1996). *Making Sense of Qualitative Data: Complementary Research Strategies*. Thousand Oaks, California: Sage

Cornelius, W.- Hollifield, J.-Martin P., (2004). *Controlling immigration: a global perspective*. Standford, CA: Standford University Press

Fairbrother P. and Rannie A. (2006). *Globalisation, State and Labour*. London: Routledge.

Fontana, A. and J. H. Frey (2002). Interview. From structured questions to negotiated text. In N. Denzin and Y. Lincoln (Eds.), *Handbook of Qualitative Research*. Thousand Oaks/London/New Dehli: Sage Publication

Glaser and Strauss (1967). *The Discovery of Grounded Theory: Strategies for Qualitative Research*. Chicago: Aldine

Glick Schiller, N. -Basch, L.-Szanton Blanc, C. (1999) 'From immigrant to transmigrant: theorizing trasnantional migration'. In L. Pries (Ed.), *Migration and transantional social spaces*. Aldershot: Ashgate

Guiradon V. and Lahav G. (2000). *A reappraisal of the state sovereignty debate: The case of migration Control. Comparative Political Studies*, 33 (2): 163-195.

Hardt M. and Negri A., (2002). *Impero. Il nuovo ordine alla prova*. Collana Storica Rizzoli: Roma.

Harvey, D. (1982). *The Limits to Capital*. New York: Oxford University Press.

Heyl, B. S. (2001). Ethnographic Interviewing. In P. Atkinson, et. al. (Eds.), *Handbook of Ethnography*. London: Sage.

Hollifield J.F. (2000). Migration and the New International Order: The missing Regime. In B. Gosh (Ed.), *Managing Migration. Time for a New International Regime*. Oxford: Oxford University Press.

Jessop, B. (2002). Liberalism, Neoliberalism, and Urban Governance: A State-Theoretical Perspective. *Antipode*, 34 (3): 452-472.

Jessop, B. (1994). Post-Fordism and the State. In A. Amin (Ed.), *Post-Fordism: A Reader*. Cambridge, MA: Blackwell.

Jessop, B. (1990). *State Theory. Putting Capitalist States in their Place.* Cambridge: Polity Press.

Jones, M. (1998). Reconstructing the local state: economic governance or social regulation? *Political Geography,* 17 959-988

Joppke C., (1998). *Challenge to the Nation States: Immigration in Western Europe and the United States*, New York, Oxford: Oxford University Press

Jordan, B. and Düvell, F. (2002). *Irregular migration: the dilemmas of transnational mobility*. Cheltenham: Edward Elgar.

Jordan, B.- Düevell F. (2003). *Migration: the Boundaries of Equality and Justice.* Oxford: Polity Press

Karakayali, S. and Tsianos, V. (2005). Mapping the Order of New Migration. Undokumentierte Arbeit und Autonomie der Migration. *Peripherie. Zeitschrift für Politik und Ökonomie in der Dritten Welt*, XXV, 97/98: 35-64.

Lahav G., (2004). *Immigration and politics in the new Europe: reinventing borders.* Cambridge: Cambridge University Press.

Lefebvre, H. (1991). *The Production of Space*, trans. Nicholson –Smith, D. Cambridge, MA: Blackwell.

Lipietz, A. (1992). *Towards a new economic order: postfordism, ecology, and democracy*. New York: Oxford University Press.

Lovering, J. (2006). *Regionalism and the Neo-Liberalisation of Governance.* (First Draft). ESRC Research Seminar Series. The rise of Multi-Level Governance and Meta Governance in an International Context, March 2-3, 2006.

Lovering J. (1998). *Globalization, Unemployment and Social Exclusion in Europe: Three Perspective on the Current Policy Debate,* International Planning Studies, Vol.3 No.1, 35-56

Massey, D.S. - Arango J. – Hugo, G. – Taylor, J.E. (1993), Theories of International Migration: A Review and Appraisal, *Population and Development Review*, 19, pp. 431-466.

Mezzadra, S. (2006). *Diritto di fuga. Migrazioni, cittadinanza, globalizzazione* (nuova edizione) Verona: Ombre Corte

Mezzadra, S. (2006a). Citizens and Subjects. A postcolonial Constitution for the European Union? *Situation,* I 2

Mitchell, K. (1997) Transnational Discourse: Bringing Geography back in. *Antipode*, 29: 2 1997 pp. 101-114

Nichols, T. and Sugur, N. (2004). *Global management and local labour: Turkish workers and the Modern Industry*. Basingstoke, Palgrave Macmillan

Ohamae, K. (1995). *The End of the Nation State.* New York: The Free Press.

Ong, A. (1999). *Flexible citizenship: the cultural logics of transnationality*. Durham: Duke University Press.

Ong, A. (1991). The Gender and Labour Politics of Postmodernity. *Annual Review of Antrhopology*, 20, 279-309.

Papadopoulos, D. -Stephenson, N. -Tsianos, V. (2008). *Escape routes. Power and subversion in the 21st century*. London: Pluto

Papastergiadis, Nikos (2000). *The Turbulence of Migration*. Polity Press: Cambridge.

Peck, J. (1996). *Workplace. The social regulation of labour markets.* New York- London: The Guildford Press

Peck J. and Tickell, A. (1992). Local modes of social regulation? Regulation theory, Thatcherism and uneven development. *Geoforum*, 23, 347-364)

Peck, J. and Tickell, A. (1994) searching for a new institutional fix: the after-Fordist crisis and the global-local disorder. In A. Amin (Ed.), *Post-Fordism: A Reader.* Cambridge, MA: Blackwell.

Potter, J. and Wetherell, M. (1995). Discourse Analysis. In J. A. Smith, R. Harre, and Van Langenhove, L. (Eds.), *Rethinking Methods in Psychology*. London: Sage.

Rigo, E. (2007). *Europa di confine. Trasformazioni della cittadinanza nell'Unione allargata*. Roma: Meltemi.

Rigo, E. (2005). *Implication of EU Enlargement for Border Management and Citizenship' in Europe*, EUI Working Papers, European University Institute, Bada Fiesolana: RSCAS Publication.

Rigo, E. (2005a). Citizenship at Europe's Borders: Some Reflections on the Postcolonial Condition of Europe in the Context of EU Enlargement. *Citizenship Studies*, 9(1), 3-22.

Sassen, S. (1997). *Informalization in advanced market economies.* Geneva: International Labour Organization.

Sassen S. (1998a). The de facto Transnationalising of immigration policy. In C. Joppke (Ed.), *Challenge to the Nation States: Immigration in Western Europe and the United States.* New York, Oxford University Press

Silverman, D. (1993). *Interpreting Qualitative Data: Methods for Analysing Talk, Text and Interaction.* London: Sage.

Silvermann, D. (2003). Analyzing Talk and Text. In N.K. Denzin and Y.S. Lincoln, (Eds.), *Collecting and Interpreting Qualitative Materials.* Thousand Oaks, California: Sage

Weiler J.H.H. (2003). *La Costituzione dell'Europa.* Bologna: Il Mulino.

-Major EU Policy documents

CEC (2001) *Commission White Paper on European governance*, COM (2001) 428; Bull. 7/8-2001

CEC (2001) *Proposal for a Council directive on the conditions of entry and residence of third-country nationals for the purpose of paid employment and self-employed economic activities* COM(2001) 386 final)

CEC (2003) *Communication from the Commission on Immigration, integration and employment,* COM 336 final

CEC (2004) *Green Paper on An EU approach to managing economic migration,* COM (2004) 811 final

CEC (2004) *Communication from the Commission Study on the links between legal and illegal immigration,* COM (2004) 412 final

EC 2005: *Council Decision on Guidelines for the Employment policies of Member States* (2005/600/EC) (New Lisbon Agenda) and Annex The Employment Guidelines (2005 to 2008)

CEC (2005) *Communication from the Commission. Policy Plan on Legal Migration* COM (2005) 669 final

CEC (2005) a *Communication from the Commission on A Common Agenda for Integration: Framework for the Integration of Third-Country Nationals in the European Union*, COM (2005) 389 final

CEC (2005) b *Communication from the Commission on Migration and Development: Some concrete orientations,* COM (2005) 390 final

CEC (2005)c *Communication From The Commission to the European Parliament, The Council The European Economic and Social Committee and the Committee of the Regions European values in the globalised world Contribution of the Commission to the October Meeting of Heads of State and Government* COM (2005) 525 final

CEC (2006) *Communication from the Commission to the Council and the European Parliament The Global Approach to Migration one year on: Towards a comprehensive European migration policy* COM (2006) 735 final.

CEC (2006) a *Commission Communication The demographic future of Europe – from challenge to opportunity*, Brussels, COM (2006) 735 final).

EC 2006 *Joint Employment Report 2006-07* Council of the European Union (6706/07)

CEC 2007 *Communication from the Commission to the Council, the European Parliament, the European Economic and Social Committee of the Regions On circular migration and mobility partnerships between the European Union and third countries* (draft, May 2007)

CEC 2007a *Directive of the European Parliament and of the Council providing for sanctions against employers of illegally staying third-country nationals*
(presented by the Commission, May 2007, draft form)

CEC 2007b *Commission staff working document Accompanying document to the Proposal for a directive of the European Parliament and of the Council providing sanctions against employers of illegally staying third-country nationals Impact assessment* [COM(2007)249]

CEC 2007c *Commission staff working paper "Measures to prevent and reduce the employment of third country nationals who are illegally staying or working in breach of their residence status* (COM 2007c) SEC(2007) 596/2

Gruber, L. (2007) *Draft Report on the Policy Plan on legal Migration Committee on Civil Liberties Justice and Home Affairs* (2006/2251/ (INI))

Klamt, E. (2005) *Report on an EU approach to managing economic migration* (COM (2004)0811 - 2005/2059(INI)) Committee on Civil Liberties, Justice and Home Affairs

Weber, M. (2005) *Draft report on the proposal for a directive of the European Parliament and of the Council on common standards and procedures in Member States for returning illegally staying third-country nationals* (COM(2005)039 2005/0167(COD); Committee on Civil Liberties, Justice and Home Affairs, Provisional 2005/0167(COD)

Five Hundred Years of Fear: Social and Medical Recognition of Huntington's disease.

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Abstract

Scientific research into genetics (and incurable diseases) is a strong feature of early 21st century medicine which has the capacity to change our perception and experience of disease. The 20th century saw major revolutions in health care such as antibiotics, chemotherapy and organ transplantation which changed our ideas about what it means to suffer from serious infection, cancer and a failing body. The 21st century opens with the exploration of the human genome, the role of proteins in the body, and the promise of stem cell transplantation, all contested areas which have the power to radically change our relationship with many diseases once again. In sociological terms, the relationship with disease is an important feature of post industrial modern life, consuming millions of pounds of investment in hospitals and medical staff, and contributing to the rise of the ageing population. The study of Huntington's disease which is known to be genetic in origin, incurable, and rare, provides a well documented example of how a disease is subject to changes in perception throughout time, space and research. New technological breakthroughs in stem cell research have served to 'move' the focus of Huntington's disease research from a mainly genetic basis to a more neurological and neuroscience basis. It is of sociological interest to observe the practice and experience of this research in clinics and laboratories, and to document the meaning of it for patients, families, scientists and clinicians. In order to understand how the research has arrived at this position, this working paper examines the social and medical recognition of Huntington's disease from the earliest known reference to it.

Introduction

Scientific research into genetics (and incurable diseases) is a strong feature of early 21st century medicine which has the capacity to change our perception and experience of disease. Those illnesses and diseases which have previously been thought of as incurable, hereditary, manifest due to quirks of fate, are becoming identifiable, testable, predictable and in some cases treatable.

The 20th century saw major revolutions in health care such as antibiotics, chemotherapy and organ transplantation which changed our ideas about what it means to suffer from serious infection, cancer and a failing body. The 21st century opens with the exploration of the human genome, the role of proteins in the body, and the promise of stem cell transplantation, all contested areas which have the power to radically change our relationship with many diseases once again.

In sociological terms, the relationship with disease is an important feature of post industrial modern life, consuming millions of pounds of investment in hospitals and medical staff, and contributing to the rise of the ageing population. The study of Huntington's disease which is known to be genetic in origin, incurable, and fortunately quite rare, provides a well documented example of how a disease is subject to changes in perception throughout time, space and research.

From the earliest known records of the disease and continuing through most of the 20th century, Huntington's disease has been feared and hidden by many families, considered to be socially unacceptable by those ignorant of its origins, and the subject of intense research by scientists and clinicians. In the case of Huntington's disease, new technological breakthroughs in stem cell research have served to 'move' the focus of disease research from a mainly genetic research basis to a more neurological and neuroscience research basis.

The latest research in the 21st century focuses on the brain processes involved in the disease with the possibility of stem cell transplantation to halt decline, if not provide a total cure. The amount and type of research currently underway is global rather than local, involving mass epidemiology, and utilises many new technological processes and experiments, with stem cells and human or animal brain material. It is of sociological interest to observe the practice and experience of this large research exercise both in clinics and laboratories, and to document the meaning of it for patients, families, scientists and clinicians. An interesting question is- how does

framing the disease as neurological instead of genetic, (with the attendant possibilities for stem cell innovation), change the relationship between the patients, families and clinicians on one hand and the disease on the other?

The project which will eventually inform the main thesis of this PhD, will concentrate on the clinic experiences of current research participants, including both patients and clinicians. Early indications are that involvement in 'big-science' research, even of a non-invasive, clinical type, potentially alters the relationship of the patient and family member to the disease in ways that may be beneficial to both the researcher and the research participant. It is also possible that this type of mass clinical research activity raises the expectations of participants and causes them to seek out particular clinics in order to benefit from the potential of more invasive research should it become available.

If successful, new processes of stem cell transplantation will change forever the relationship of families and society to this disease, perhaps in a similar way to the changes paved by antibiotic use, cancer treatments and organ transplants. Current research has already contributed to a new understanding of transmission and spread of the disease from healthy individuals who have a particular combination of a repeated sequence in their DNA (Harper: 2002). In order to appreciate the many different ways in which the disease has been regarded over time, and how research has accumulated to date, this working paper sets out the history of social and medical recognition of the disease.

The disease known as HD- Huntington's Disease.

This working paper will outline the history of social and medical recognition of the disease, how the disease has spread, major family studies, disease epidemiology and the genetic basis of the disease.

HD or Huntington's disease (previously known as Huntington's chorea), remains one of the most feared and devastating of genetic diseases. It affects successive generations of whole families, making no distinction between age, gender, education, or personality. It is a neurodegenerative disease which progresses in several stages, and becomes visible due to involuntary twitching and jerking movements of the face, limbs and body (usually leading to complete disability), loss of cognition and insight and often dementia. Death often occurs approximately fifteen to twenty years after onset and is often caused by complications of the effects of Huntington's disease.

The disease usually affects adults in middle life, although it is known there is some variability in age of onset and severity of symptoms (Kremer: 2002, cited in Bates et al, 2002, p.29). Huntington's disease is dominantly inherited, which means that every child of an affected adult has a one in two chance of inheriting the gene and developing the disease themselves in adulthood, and importantly, of potentially passing the gene on to their own children who then run the risk of being similarly affected. Despite the disease being recognised for well over a century, there is as yet no treatment or cure, although there are now some drug treatments that can alleviate some of the symptoms.

It is now known that the genetic mutation for Huntington's disease causes different behaviour of 'huntingtin' protein in the brain of the affected person. The proteins become attached to brain cells in an irreversible process, leading to cell atrophy and eventual cell death, shrinking the mass of the caudate nucleus and many other areas of the brain (Gutekunst et al:2002). Eventually, the build up of proteins prevents the correct neuronal activity in areas of the brain responsible for body movement, cognition and insight. Although proteins often attach to brain cells in everyone, this is usually a reasonably spontaneous reversible process. It is not yet known why the process in Huntington's disease becomes irreversible during middle life. Some of the current research is concentrated on the interruption and arrest of this attaching process which is believed to begin before there is any external physical sign of the disease. However, it is considered there may be very subtle cognitive changes which can be detected by careful observation and research (Kipps et al: 2005). In the next section I will discuss the history of the disease recognition and classification.

History of the disease name or classification

The idea that excessive body movements, which characterise the disease, are linked to problems in the central nervous system was first put forward by Paracelsus (1493-1541, cited in Hayden, 1981). He introduced the name 'chorea' to medical literature, derived from Latin 'choreus' which alludes to dancing. The three hundred years between Paracelsus and George Huntington's account in 1872 which gave the disease its name, saw much nosological confusion about different illnesses that had choreic features, but these were attributable to many different causes and circumstances of onset. 'Chorea' is now widely used as a medical term, to mean involuntary movements, but not all 'chorea' described in early medical literature, was due to

Huntington's disease. Some diseases may have been Huntington's, however from their descriptions, and the knowledge we have today, this is by no means certain.

References are made to 'St.Vitus dance' as an alternative name for Huntington's disease, but Hayden (1981) claims that this is an error made by Thomas Sydenham in 1686. Sydenham was a highly esteemed English physician sometimes known as 'the father of chorea' due to his work in accurately describing the symptoms and signs of choreic type diseases. He also identified the condition as 'St Vitus dance', an unfortunate link which has persisted for over 300 years. This term is sometimes used by lay people in connection with Huntington's disease, but it is widely accepted in clinical practice that 'St Vitus dance' refers to the 'dancing manias' of the Middle Ages which were eventually attributed largely to hysteria. The condition(s) described by Sydenham also became known variously as 'Sydenham's chorea', 'chorea anglorum' (to honour the contributions to understanding made by English physicians), and 'chorea minor' as distinguished from 'chorea major', which was the 'dancing mania' previously referred to.

The first reference to an inherited form of the adult onset disease came from Elliotson an Englishman, in the Lancet of 1832 (Stevens: 1972). However, Harper (2002) claims that it is unclear whether or not Elliotson was referring to Huntington's disease, and cites Waters account in 1842 as being a more trustworthy first written account. This appeared in the form of a letter to the editor in 'Practice of Medicine', vol 2, (Dunglison:1842, cited in Harper, 1991).

Two other early references to HD came from Lund, a Norwegian in 1860, and Lyon in the USA in 1863, are also discussed by Harper. The Lund account appears to have been mostly accurate but was disadvantaged by being written in Norwegian and not translated outside Norway. An additional disadvantage was that it was confused with Parkinson's disease by some commentators. The description by Lyon is not completely credible an account of HD, due to striking differences in the nature and time of onset of chorea, which according to Harper suggest a diagnosis of benign familial chorea rather than HD (2002).

The first accurate description of a disease or chorea, as an inherited adult onset disease, and a separate entity from other disorders that caused involuntary movements, was made by George Huntington in 1872. He gave a paper at the Meigs and Mason Academy of Medicine at Middleport, Ohio, on 15 February 1872, and published it two months later in the journal 'The Medical and Surgical Reporter'. The second part of the paper gave such authentic clinical detail,

that it is considered to be as relevant and accurate a description today as it was at the time it was first published (Harper: 2002).

Huntington published only this single paper on the subject of inherited chorea, but his observational work and that of his father and grandfather amounted to 78 years of medical practice spanning their three generations in Long Island, USA. Due to the concise and accurate nature of George Huntington's disease description, his work was translated into German and his name began to be associated with the disease in different parts of the world. The use of the word 'disease' rather than 'chorea' appears to have gained more acceptance in the late twentieth century, due to the recognition that physical rigidity is also a feature of the disease, and that 'chorea' (indicating movement) is not always an entirely accurate description (Hayden: 1981). Despite the move away from using eponyms to describe medical disorders, there is no international consensus on an accurate, non-eponymous alternative name for the disease, so for the time being Huntington's chorea, or disease, remains the term of reference.

The following table is useful because it shows most of the major steps in recognition of the disease from the early nineteenth century.

Table 1.1 Landmarks in the history of Huntington's disease (reproduced in full from Hayden, 1981, p.10)

1832	John Elliotson, the English physician, first mentioned the possible influence of heredity on adult chorea
1841	The Rev. Dr. Waters (1816-1892) described adults with hereditary chorea in the New York area in a letter to his Professor of Medicine
1842	Charles Gorman submitted a dissertation 'On a form of Chorea Vulgarly called Magrums'
1858	George Huntington's first exposure to affected patients whilst accompanying his father on his rounds
1860	Johan Christian Lund, district physician in Norway, described chorea St. Vitus which 'recurs as a hereditary disease in Saetersdal'
1863	Irving Lyon submitted article on chorea to American Medical Times, describing affected families in Connecticut.
1872	George Huntington's definitive description of the disease
	George Huntington's article abstracted into German

1887	Huber first used eponym Huntington's chorea to describe hereditary chorea
1888-1895	Eponym adopted by different authors in France, Italy, England, and USA
1908	First monograph on Huntington's chorea (edited by Browning), citing over 200 references
1909	George Huntington addresses the New York neurological society
1916	Death of George Huntington
1967	The first meeting of the World Federation of Neurology Subgroup on Huntington's Chorea in Montreal, Canada. Meetings subsequently held biennially
1972	Centennial Symposium on Huntington's chorea in Columbus, Ohio, USA, in commemoration of Huntington's original contribution
1977	The Commission for the Control of Huntington's disease in the USA reports to Congress and the President
1978	Second International Symposium on Huntington's disease held in San Diego, California, USA

Hayden published his monograph in 1981, long before the gene for Huntington's disease had been located, and therefore there are several further important events to be added to a table of this nature. These are reproduced here from Harper (2002, p.18) together with other landmarks in the study of HD cited by Harper but not by Hayden. These include events of clinical and historical importance prior to 1981. Taken together, the tables provide a more complete picture of the development of interest in the disease. It is noted that the authors named their tables slightly differently; Hayden uses 'history', whereas Harper prefers 'study'.
(see Table 1.2 below)

Table 1.2 Landmarks in the study of HD *(reproduced in part from Harper, 2002, p.18)*

1888	Juvenile HD clearly described (Hoffman)
1908	Mendelian dominant inheritance recognised
1934	Systematic study of inheritance (Bell)
1958-59	First detailed genetic-epidemiological survey in specific region (Michigan)
1967	Committee to Combat Huntington's Disease is formed
1983	Localisation of HD gene on chromosome 4
1987	First applications of DNA markers in prediction
1993	Isolation of HD gene; identification of mutation as expanded CAG repeat
1994	Polyglutamine repeat recognised as important in neuronal pathology (Perutz)
1996	Neurological phenotype produced in transgenic mouse with expanded HD repeat
1997	Neuronal inclusions recognised in transgenic mouse and human HD brain

The spread of Huntington's disease throughout the world

The disease spread can be closely linked to population movements, especially from mainland Europe over the last three hundred years, and interest has often focussed on relatively new countries such as the United States, Canada and Australia. The tracing of ancestors from Europe has thrown light on some of the disease incidence in these countries (Harper: 2002)

There is some controversy or dispute over the spread of HD from the UK to the United States, with some accounts claiming that individual named immigrants from Bure in Suffolk, carried the disease to New England in the early seventeenth century (Vessie: 1932, Critchley: 1934, cited in Harper, 2002, p.171). However, although the geographical links to migrants from East Anglia is considered to be sound, the exact source of the disease from one or two named individuals has never been demonstrated satisfactorily (Caro:1979 unpublished thesis, cited in Harper, 2002 p.171).

It is speculated that the appearance of the disease in some of the Pacific Islands, may be due to transmission via the nineteenth century whaling industry (Scrimgeour & Opitz: 1983). This may

be possible to verify with the use of recent technology that allows for the analysis of molecular haplotypes, but as yet this research has not been conducted. Harper asserts that this would be an interesting project as it would demonstrate the lasting effects that genetic disorders create in a population long after the original mechanism for transmission has ceased (2002). Bona-fide reports of the disease persisted throughout the late nineteenth and the early years of the twentieth century, covering most of Europe from Russia in the East to the UK in the west, USA and Canada, Cuba, Brazil, and Argentina in the Americas, plus Australia, up to 1904 (based on Bruyn et al:1974, cited in Harper, 2002, from Table 1.1 p12).

The spread of HD within countries can be linked more strongly to internal migration (Harper: 2002), and in the United States and Australia this can be seen in the patterns of settlements made by people already in the country, created by their geographical movements to find work or farm land. 'More settled' countries in Europe including the UK, also show patterns of spread from internal migration, and 'foreign migration' accounts for much less of the spread of the gene. This was particularly evident in the South Wales HD study by Walker et al (1981), where 4 percent of families originated outside Britain, and 43 percent were from outside Wales. This is no doubt linked to the influx of workers to the Welsh industries from other parts of the UK at the time of the Industrial Revolution. During this time, rural areas were hard pressed to support large families, and many came to the Welsh valleys seeking better paid work in the iron industry and later, in coal mining. The South Wales study found that there was proportionally a greater migration of HD ancestors from South West England to industrial South Wales than from any other area of Wales or the UK .

In addition to the spread of the disease through population movement, an additional feature can sometimes be seen; this is the geographical concentration of HD affected people and their at-risk relatives where there has been very little later migration (Harper: 2002). Harper gives the example of the sometimes static nature of established kindreds, where a large family in South Wales was descended from a single immigrant from outside Wales.

"Of the 55 known and 177 at risk descendants, only 24 had moved further than a 10 kilometre distance from the original point of settlement 120 years ago" (Harper:2002).

This lack of migration by affected families has made studying these affected populations slightly easier, and there are well-documented examples including Venezuela, North Sweden, Tasmania, Moray Firth in Scotland, Mauritius, and the South Wales population, which is the subject of this

thesis. There were many more studies carried out worldwide, and interestingly, the recorded incidence of the disease in some areas of low population gives rise to high prevalence rates per million. These must be treated with caution because there was no standardisation of methods in early surveys, for example- the English Counties study (Critchley :1934), and a London study (Minski & Guttman:1938). Therefore not all studies have the same amount of importance attached to them, or can be considered accurate enough to be used for comparisons (Hayden:1981). Today, Huntington's disease is found in most developed countries, although it is thought to be less prevalent in sub-Saharan African and Asian populations, and is less prevalent in Finland and Japan.

Family studies of the disease

Early family studies of the disease were carried out in the USA (Davenport and Muncey: 1916), the United Kingdom (Julia Bell:1932), Wales (Spillane & Phillips:1937) and Germany (Panse:1942). Unfortunately, some of the studies showed biases towards eugenic policies and contributed to the mistrust of genetics as a useful site of medical study.

Davenport and Muncey (1916) claimed to have traced almost a thousand cases from Long Island and parts of Connecticut and Massachusetts, to the migration of three English brothers in the seventeenth century. It was clear from the views expressed in the paper that Davenport held strong opinions on the potential for the screening of immigrants.

"All these evils in our study trace back to some half-dozen individuals including three brothers, who migrated to this country during the 17th century. Had these half-dozen individuals been kept out of this country much of the misery might have been saved." (Davenport and Muncey:1916, p.215).

Davenport's later association with German race-hygienists is therefore perhaps unsurprising (Harper: 2002). Harper (1991a) makes the point that it is unlikely that a policy of refusing immigration to individuals would have been effective in preventing HD in America. Population studies have since shown the multiple origins of HD and involved a variety of ethnic groups (Harper:1991a, cited in Harper 1992, p461). However, the Davenport and Muncey study was important in several other ways. They identified what they termed 'biotypes' within the main disease population they studied, where either tremors or mental deterioration were present individually, and other 'biotypes' where the chorea did not progress, or the onset was juvenile.

They correctly identified the dominant inheritance pattern, and also referred to the 'law of anticipation' in relation to the age of onset, but concluded that this was illusory, due to not being able to compare on the same basis for onset in grandparents and grandchildren. Anticipation is a phenomenon whereby the symptoms of the genetic disorder become apparent at an earlier age as it is passed on to the next generation. In most cases, an increase of severity of symptoms is also noted. It is common in trinucleotide repeat disorders like Huntington's disease, myotonic dystrophy and fragile x syndrome, where triplet repeat mutations in DNA are implicated. Although knowledge of the trinucleotide repeat responsible for Huntington's came much later in the twentieth century than the Davenport and Muncey study, in 1916 they had accurately identified anticipation as an important feature of the disease manifestation.

Davenport and Muncey (1916) also considered that there was a higher than normal incidence of nervous troubles in the three thousand relatives of the affected population, leading them to suggest that chorea occurs in families characterised by mental troubles. This is in stark contrast to the more widely held view today, that due to the existence of the disease in families, people in those families suffer quite understandable anxieties and mental anguish.

The Davenport and Muncey study also concluded that the families recognised the hereditary nature of the disease which did not skip generations, "nevertheless, there is no clear evidence that persons belonging to the choreic lines voluntarily abstain to any marked degree from, or are selected against, in marriage".(Davenport and Muncey: 1916). This comment on the social situation of people in families at risk of Huntington's disease speaks volumes about the prevailing attitudes of the time on the importance of marriage as a contract, and the pressure to reproduce, providing future generations of healthy Americans to populate a relatively new country. Despite their rather unsympathetic account of the families affected by Huntington's disease, the Davenport and Muncey contribution of detailed data concerning age of onset, variations between families, anticipation and biases, types of mental involvement, and varieties of motor disturbances, is considered to be the extremely solid foundation of much future work.

The 1934 account of the disease by Julia Bell was a systematic study of inheritance. This British study was sympathetic and perhaps ahead of its time in the way Bell viewed the situations of families at risk of Huntington's disease.

"The almost continuous anxiety of unaffected members of these families over so long a period must be a great strain and handicap, even if they remain free from disquieting symptoms; it is thus of urgent importance that some means should be sought by which the immunity of an individual could be predicted early in life, both from the point of view of relief to those who carry no liability to the disease and as an indication to others that they should abstain from parenthood. No facts in the clinical histories of patients provide definite guidance in this matter prior to the onset of symptoms, but the development of the science of genetics may at some future date enable us to obtain information concerning the inherent characteristics in such cases." (Bell:1934)

Interestingly, Bell is not referred to in the 1937 account of 'Huntington's Chorea in South Wales' (Spillane & Phillips:1937), and a paragraph entitled 'The Hereditary Process' mentions only the 1916 Davenport and Muncey study. This could be because the focus of the Spillane and Phillips' South Wales study was primarily concerned with how the disease was manifesting in the Welsh population, but it is not unfeasible that the work of Bell was disregarded because she was a woman. Harper (2006) refers to several examples in the early 20th century where the scientific work of women in genetics was overlooked in favour of male colleagues working in the same laboratory.

In the Welsh study, Spillane and Phillips examined six family groups; all but one originated in Wales. Their research looked at disease incidence, symptomology, mode of inheritance, and 'the constitution, ability, and state of the nervous system in all members, together with an account of their attainments in life'. The authors described it as 'a detailed genealogical and sociological study of the families concerned' (Spillane and Phillips: 1937). It is noteworthy that this study also openly gave the view that there should be legislation to prevent individuals at risk from marrying and having children.

"*Perhaps with repeated advice and education, some would voluntarily abstain from marriage, but the majority would no doubt be prepared to accept the even chance that nature offers them. We are left with the conclusion that only legislative measures will eventually succeed in eradicating the disease."* (Spillane and Phillips:1937)

The 1942 study by Panse, is controversial because he reported all his patients and families affected with Huntington's to the German authorities, even though he was probably aware that this meant compulsory sterilisation and possibly death for most of them (Muller-Hill:1988). Panse, known as a prominent expert in the study of HD and also a member of the Nazi party, was closely involved in drawing up the race-hygiene laws in Germany (Muller-Hill:1988). The

Nazis in 1933 had invoked eugenics policy via the 'Law for the Prevention of Hereditarily Diseased Progeny' as part of their bid to strengthen the Germanic race. This was an open policy on compulsory sterilisation which was carried out by a series of genetic health courts each led by a lawyer and two doctors (Harper:1992). The policy culminated in the Holocaust, but its beginnings were aimed at creating a pure Aryan race by removing all types of 'defectives' from society, including mentally and physically handicapped individuals. Panse recorded that he had reported "all choreic cases, and moreover all suspicious cases and finally all not yet choreic sibs and offspring as being at risk to the health authorities…." There is no doubt that killings took place of all types of people considered to be defective or 'risky', despite the fact that the 1933 act provided only for affected persons to be sterilised compulsorily. Most of the 400,000 sterilisations were carried out before the beginning of the Second World War and included both males and females. Historians claim that this was a manifestation on the 'collective plane' of Hitler's personal exaggerated fears of disease (Bidiss:1997). However, the study by Panse of over a million people, is thought to be one of the first thorough and accurate pieces of work, which gave prevalence estimates which closely agreed with much later post-war analyses of three to five affected people per hundred thousand (Harper: 2002).

Thus, genetics for a time became synonymous with the worst excesses of eugenics in Germany where it was designed to rid the country of feeblemindedness and psychosis (Mazumdar : 2002). It was some time after the Second World War that the 'new genetics' was considered by some to be a sufficiently different concept to the previously tainted model, although Mazumdar (2002) considers the gradual post-war change to be more of a shift away from Mendelism. After the war, Panse was re-appointed to a senior academic post with the full co-operation of the Allies. Bidiss (1997) states clearly : " Kater [1989] provides ample evidence that physicians became Nazified more thoroughly and much sooner than any other profession". This may provide a partial explanation of why Panse was acquitted of crimes against humanity and continued in academic study - there were far too many physicians who had acted in a similar way to usefully pursue them all, plus the Allies saw value in their continuing work in genetics.

Epidemiology of the disease

Harper puts forward the view that traditional epidemiology is not completely successful in gaining information about the prevalence of this disease for several reasons (Harper: 2002). Many chronic neurological disorders such as multiple sclerosis, stroke and motor neurone disease, have successfully utilised an epidemiological approach in hypotheses about causation,

frequency and variation. The situation with Huntington's disease is different because since the gene discovery in 1992, it is evident that a specific gene underpins the manifestation of the disease in the population. Therefore, the molecular details of the disease will be more crucial than epidemiology in determining pathogenesis, that is, the origins and effects of the disease (Harper:2002).

Most of the detailed information known about the frequency in individual national populations has come from detailed family studies carried out with painstaking attention to detail by generations of interested clinicians as described above. Harper also carried out detailed family studies as part of his work in establishing the Institute of Medical Genetics in Cardiff from the early 1970's, and also as part of the Huntington's Disease Collaborative Research Group in the 1980's. He found that with careful information giving and non-directive genetic counselling, there appeared to be a reduction in the birth rate for families at risk (Harper et al:1981, Carter et al:1983). This was interesting because of the observed higher than average fertility in families where Huntington's was present (Walker et al:1983). A later paper by Harper concluded that prediction and testing was a still a long way ahead, but in the meantime, clinicians could still provide a very valuable service by providing families with good communication of information and support (Harper: 1985).

Most of the family studies were carried out prior to the isolation of the HD gene in 1993. Harper asserts that the genetic discovery means that the family studies can be interpreted differently since that time (2002). For example, there are often different genetic mutations giving rise to the same disease, such as in cystic fibrosis, where there is one frequent mutation and several rare ones. In Huntington's disease it has been known since linkage data became available that there is 'only a single major genetic locus involved', (Gusella et al:1983, Conneally et al: 1989, cited in Harper, 2002 p 186) and it was considered that the number of different mutations would be small. (Linkage data is explained in the next section, "Genetics of Huntington's disease".) The discovery of the instability of the trinucleotide repeat of the protein chain 'CAG' on chromosome 4 and that the same mutational mechanism is involved in all cases of HD, appears to have quashed the original hypothesis that several different mutations may be involved, and that large family studies would assist in revealing the nature of these. The next section will discuss what is known about the CAG repeat sequence in Huntington's disease after a short introduction to the basics of Mendelian genetics.

Genetics of Huntington's disease.

Basic genetics can show patterns of inheritance of plants, animals and humans, and the theory associated with simple inheritance was published in 1865 by Gregor Mendel. Mendel was an Austrian monk who ran a series of breeding experiments with peas in the 1850's, to discover the characteristics which were passed on from one generation to another. His findings were not recognised during his lifetime, it was 1900 before his work became accepted as solid scientific fact.

A contemporary description today would explain that in human reproduction our chromosomes and genes are set out in 23 pairs.

The chromosomes are inherited from our parents, one from each parent in each pair of chromosomes. Each chromosome is a string-like structure of tightly packed molecules of DNA and is described as having a 'long arm' and a 'short arm'. The DNA consists of proteins which are labelled A, C, G and T arranged in different sequences along the structure, usually referred to as the coding of the DNA. Humans share much of this coding sequence with animals; there are fewer differences than were anticipated at the beginning of the Human Genome Project, which effectively discovered the whole of the human DNA sequence in 2001. Whatever is contained in the chromosomes, controls what features we have inherited from our parents. This includes such things as hair and eye colour, but also the *potential* for height, weight and many other things. What is not completely understood is the interaction between genes and environment which may alter the effect that genes actually create in the individual. It is important to recognise that in many cases it is impossible to say that a particular gene is responsible for a characteristic or type of behaviour, because of the little understood interactions between different genes, and additionally between genes and environment. Unfortunately in Huntington's disease, there does not appear to be much evidence of a mitigating environmental effect to the extent that a person with an extended CAG sequence within the range for developing HD, will not develop the disease. However, there is variation in the time of onset of HD, and the fine detail of how someone is affected with different emphases on movement, cognition, behaviour and insight.

Every species has its own number of chromosomes per cell, known as the diploid number, humans have 46 of these arranged in pairs. There are 23 homologous chromosome pairs, 22 pairs of autosomal chromosomes and two distinct sex chromosomes X and Y. It is thought that

there are at least 20-25,000 different genes in the human genome. Each chromosomal pair has the same genes, although it is unlikely that homologous genes (sharing the same position or function) from each parent will be identical in sequence.

There are variants possible for individual genes known as alleles, for example there is an allele for blue eyes, a different one for brown eyes, and so on. When one allele causes expression of a feature over the other it is said to be dominant. The allele for brown eyes is dominant, so even if a child inherits one brown eye allele and one blue eye allele, the child will have brown eyes. The allele for Huntington's disease is autosomal dominant, which means that if one copy is inherited from a parent, the disease will develop in the offspring. Some conditions are known as autosomal recessive, which means that allele copies from both parents must be inherited for a disease to be expressed in the offspring. Examples of recessive diseases inherited in this way are cystic fibrosis (CF) and phenylketonuria (PKU).

The dominant inheritance pattern of Huntington's disease had been noticed by George Huntington who described it as follows:

"One or more of the offspring almost invariably suffer from the disease, if they live to old age. But if by any chance these children go through life without it, the thread is broken and the grandchildren and great grandchildren of the original shakers may rest assured that they are free from the disease." (Huntington:1872)

Huntington's description of genetic dominance is uncannily accurate and commensurate with the contemporary criteria as applied to HD, even though Mendel's work was not particularly well known at the time. The following table gives the current criteria for autosomal dominance as applied to HD. (See Table 1.3 below)

Table 1.3 Criteria for autosomal dominant inheritance as applied to HD

Equal incidence in both sexes.
Equal transmission by both sexes (but differential effect of affected parent on age of onset).
50% of offspring of an affected person also become affected (but only by old age)
No transmission of the disorder by those offspring remaining free from HD.

(*From Harper & Jones:2002, p114*).

Genetic and Molecular studies of Huntington's disease

As already discussed in a previous section of this chapter concerning the history of the disease classification, the hereditary nature of HD was apparent even in very early descriptions of chorea, and was distinguishable from other forms of choreic disease. Also earlier in this chapter, reference was made to the twentieth century re-emergence of Mendel's work on inheritance several years after his death. Following the re-emergence of his work, scientists made efforts to find examples of this in human disease, and it was soon apparent that HD was one such example. The early documenting of this came from Jelliffe (1908), Punnett (1908) and Davenport (1911), then Davenport and Muncey (1916). The family study by Julia Bell has already been mentioned in the context of its empathy with affected families, but the systematic, quantitative analysis of her work gave a detailed foundation for future studies (Harper and Jones:2002). A great deal of work in preceding years led to the discovery and isolation of the gene for HD in 1993, and Harper and Jones describe it as follows:

… *"new discoveries did not appear from nowhere, but grew in a systematic, if at times slow and not entirely predictable, manner from earlier studies. It is important not to forget these earlier steps, even if their significance has largely been superseded by the later work for which they provided essential foundations."*
(*Harper and Jones:2002, p.116-7*)

The information about the structure on the single gene which gives rise to this devastating disorder was discovered in 1993 (Huntington's Disease Collaborative Research Group, 1993). It was the culmination of over twenty years of research by various groups worldwide (Lindstrom et al: 1973, Brackenridge et al: 1978, Pericak-Vance et al:1978, Hodge et al: 1980, Volkers et al: 1980, all cited in Harper and Jones:, 2002, p.117). The HD gene discovery was the result of a classic gene mapping study, during which many techniques were developed which are now widely used in other disease mapping research studies. But before mapping could be accomplished, genetic linkage needed to be established. Early genetic studies of simple organisms such as Drosophila (fruit fly) had identified genetic linkage as being of importance to gene-mapping (Harper & Jones: 2002). Linkage means that two genes are situated so closely together on the chromosomes that they are co-inherited in most cases. In 1983, the Huntington's gene was located somewhere on the short arm of chromosome 4 by using a marker named G8 (Gusella et al: 1983). This discovery had several implications immediately, firstly it allowed the development of positional cloning to establish the exact location and typology of the gene, and furthermore, it enabled the possibility of pre-symptomatic testing of individuals at risk of HD.

There was also the opportunity to begin pre-natal testing of at risk pregnancies (Harper and Jones 2002). Gusella and her colleagues had only decided to try to map the gene one year before they discovered it, which in research terms was a very speedy result, and this encouraged the continuation of the work to pinpoint the details. The large family studies became increasingly valuable as they were utilised to intensely narrow the search area of the chromosome (Harper and Jones 2002). The co-operation and collaboration of the known HD population was vital to the later success of the exact location of the gene, as they were tested for possible mutations in the critical area. Several genes in the region were cloned because they had trinucleotide repeat sequences. Trinucleotides are combination sequences of nucleotides found in molecules of nucleic acid in both DNA and RNA, each indicated by one of the letters, A, C, G, T. These are indicative: A=adenine, C= cytosine, G = guanine, T = thymine. In DNA, A & T are always paired, G & C are always paired. The indication for this being a possible factor came from earlier cloning of disease causing genes in the 1990's, which also had similar repeat sequences (fragile x syndrome and myotonic dystrophy for example). After a few false starts, the 'interesting transcript 15' (IT15) gene was found to be the missing piece of the puzzle.

"*Several repeats were analysed that proved not to be expanded in HD and then a gene in the critical region, the inspiringly named 'interesting transcript 15' (IT15), proved to contain a CAG repeat in its first exon that was expanded in HD cases (HD Collaborative Research Group 1993). This landmark event was the successful end of a 10 year cloning effort, involving six research groups, four in the USA and two in the UK- a truly international collaboration." (Harper:2002)*

In an unaffected person, the Huntington gene is still present on chromosome 4 but the number of CAG repeats will vary up to approximately 34. In people with the mutation for Huntington's disease, the repeated CAG sequence will be in greater numbers than this, possibly from 36 repeats up to 50 or more. This is known as an abnormally long trinucleotide repeat. It is generally agreed by clinicians that a result of 39 repeats in a person showing symptoms of HD is likely to mean that they have the disease (Harper and Jones:2002). Other diseases have much greater numbers of repeats creating pathogenesis, for example fragile x syndrome has over 230 repeats of the trinucleotide sequence CGG, and spinocerebellar ataxia type 8 has between 110 and 250 repeats of CTG.

The knowledge that the area of DNA containing the gene for Huntington's disease contains an *unstable* sequence of the trinucleotide repeat CAG, and that all HD wherever in the world, is 'due

to the same mutational mechanism and show an identical, although variable in extent, CAG repeat expansion' (Harper: 2002), has major implications for the determination of origins. The range of CAG repeats in a stable, normal population will contain some individuals in the 'between range' (34 repeats of CAG) and may account for the eventual appearance of disease when, after many generations the clinically significant levels of CAG repeats are reached (Harper: 2002). The number of generations required for this to happen due to instability of the repeat is not known, neither is the combination of intermediate CAG repeat ranges which give rise to the appearance of HD in the next generation, nor the mechanism by which the process occurs. However, it has been found that in every person who has no particular family history and who develops HD, their unaffected parent(s) are found to carry CAG repeats in the intermediate range (Harper: 2002). It is the *instability* of the CAG repeat sequence length during foetal cell division which determines whether or not someone potentially develops the disease, and not simply inheriting an exact copy of the allele, although if the parent has an expanded repeat, any child has a 50% chance of inheriting that copy of the allele and becoming affected by the disease. This alters the premise that all HD worldwide originated from one or two individuals, and that the spread was entirely due to migration of affected individuals. We must therefore revise our view of the concept of mutation, and thus the spread or otherwise in populations (Harper: 2002).

Conclusion

The many and varied explanations of Huntington's disease over at least two centuries, have contributed to much of the current understanding but have yet to find a cure for this devastating disease. Unfortunately in Huntington's disease, there does not appear to be much evidence of a mitigating environmental effect to the extent that a person with an extended CAG sequence within the range for developing HD, will not develop the disease. However, there is variation in the time or age of onset, the first symptoms, and the fine detail of how different individuals are affected in terms of the speed of progression, and the order and extent of loss of motor skills, loss of cognition, and loss of insight.

Current research is focussed on mass global epidemiology, collection of tissue samples, and the study of the brain processes involved in decline of the individuals' cognition, movement, and insight. This has societal implications for the global collection and distribution of data both in terms of clinical records and human tissue samples, and for the motivation of human research participants. There are also issues concerning the work of the clinic and the normalisation of research practice into disease management, and whether this occurs purposively, spontaneously

or as a matter of coincidence. Nevertheless there exists an identifiable shift in the boundaries between clinical management and clinical research, which has been termed 'blurring' (Ponder et al: 2008) and 'routinisation' (Foster et al: 2006). In sociological terms, this may be illustrative of a scientific tension between disease management and research, which would have wide ranging implications for the ethics and practice of clinical management of this disease.

Bibliography

Bates, G. Harper, P. & Jones, L. (2002) *Huntington's Disease.* (Third Edition). Oxford Monographs on Medical Genetics, OUP, Oxford.

Bell, J. (1934) Huntington's chorea. In *Treasury of Human Inheritance* (ed Fisher, R.V., vol iv, part 1, pp 1-67, Cambridge University Press.

Bidiss, M (1996) Disease and Dictatorship: the case of Hitler's Reich. *Journal of the Royal Society of Medicine,* vol 90, pp 342-346.

Brackenridge, C.J., Case, J., Chin, E., Prospect, D.N., Teltscher, B., & Wallace, D.C. (1978) A linkage study of the loci for Huntington's disease and some common polymorphic markers. *Ann. Human Genetics* 48, pp 203-211.

Bruyn, G.W. (1968) Huntington's chorea. Historical, clinical and laboratory synopsis. In *Handbook of Neurology* (eds Vinken, P.J. & Bruyn, G.W.) v 16, pp 298-378, Elsevier, Amsterdam.

Bruyn, G.W., Baro, F., & Myrianthopoulos, N.C. (1974) *A centennial bibliography of Huntington's chorea 1872-1972.* Niijhoff, The Hague.

Caro, A. (1977) A genetic problem in East Anglia. Huntington's chorea. PhD thesis, University of East Anglia.

Carter, C.O., Evans, K.A., Baraitser, M. (1983) Effect of genetic counselling on the prevalence of Huntington's chorea. *BMJ*, vol 286, pp 281-283.

Conneally, P.M., Haines, J., Tanzi, R. *et al* (1989) No evidence of linkage heterogeneity between Huntington disease (HD) and G8 (D4310). *Genomics* 5, pp 304-308.

Critchley, M. (1934) Huntington's chorea and East Anglia. *Journal of State Medicine*, 42: pp 525-587.

Davenport, C.B. (1911) *Heredity in relation to eugenics*. New York, (republ. Arno Press 1972)

Davenport, C.B., & Muncey, E.B. (1916) Huntington's chorea in relation to heredity and eugenics. *Eugenics Record Office Bulletin.,* 17:195-222

Dunglison, R. (1842) *Practice of medicine*, 1st edition, Philadelphia, Lee and Blanchard, cited in Harper, P.S. (Editor) (1991) *Huntington's Disease. Major Problems in Neurology no. 22,* W.B.Saunders, London, Philadelphia, Toronto.

Forno, L. & Norville, R. (1979) Ultrastructure of the neostriatum in Huntington's and Parkinson's disease. *Advances in Neurology, 23, pp123-135*.

Foster, M.W., Royal, C.D.M., Sharp, R.R. (2006) The routinisation of genomics and genetics: implications for ethical practice. *Journal of Medical Ethics*, 2006, 32, pp 635-638. Downloaded from jme.bmj.com 29 /11/2006.
Gusella, J., Wexler, N.S., Conneally, P.M., Naylor, S.L., Anderson, M.A., Tanzi, R.E., Watkins, P.C.,

Ottina, K., Wallace, M.R., Sakaguchi, A.Y., Young, A.B., Shoulson, I., Bonilla, E., and Martin, J.B. (1983) A polymorphic DNA marker genetically linked to Huntington's disease. *Nature, 306, pp 234-238*

Gutekunst, C.A., Norflus, F., Hersch, S. (2002) The neuropathology of Huntington's disease. In

Harper, P.S. (1985) The Prevention of Huntington's Chorea.. The Milroy Lecture 1985, *Journal of the Royal College of Physicians of London,* vol 20 no 1 Jan 1986.

Harper, P.S. (Editor) (1991) *Huntington's Disease. Major Problems in Neurology no. 22*, W.B.Saunders, London, Philadelphia, Toronto.

Harper, P.S. (2002) Huntington's disease: a historical background. In Bates, G., Harper, P.S., &

Jones, L. (2002) *Huntington's disease*.

Harper, P.S. & Jones, L. (2002) Huntington's disease: genetic and molecular studies. In Bates, Harper & Jones (2002) *Huntington's disease*.

Harper, P.S., Tyler, A., Smith, S., Jones, P., Newcombe, R.G., McBroom, V. (1981). Decline in the predicted incidence of Huntington's chorea associated with systematic genetic counselling and family support. *The Lancet*, Aug 22, 1981 pp 411-413.

Harper, P.S., Walker, D.A., Tyler, Audrey., Newcombe, R.G., Davies, Kathleen., (1979) Huntington's chorea. The basis for long term prevention. *The Lancet, Aug 18 1979, pp 346-349*.

Harper, P.S., Youngman, S. Anderson, M.A., Sarfarazi, M., Quarrell, O., Tanzi, R., Shaw, D., Wallace, P. Conneally, P.M. Gusella, J.F. (1985) Genetic linkage between Huntington's disease and the DNA polymorphism G8 in South Wales families. *Journal of Medical Genetics, vol 22, pp 447-450*.

Hayden, M.R. (1981) *Huntington's chorea*. Berlin, Springer.

Hodge, S.E., Spence, M.A., Crandall, B.F., Sparkes, R.S., Sparkes, M.C., Crist, M., & Tideman, S. (1980) Huntington disease: linkage analysis with age-of-onset corrections. *AM.J.Med.Genet*. 5, pp 247-254.

Huntington, G. (1872) On chorea. *Medical and Surgical Reporter* 26: pp 320-321

Jelliffe, S.E. (1908) A contribution to the history of Huntington's chorea: a preliminary report. *Neurographs* 1: pp 116-124. In Bates, Harper & Jones, (2002)

Kater, M.H. (1989) *Doctors under Hitler*. Chapel Hill, University of North Carolina Press

Kipps, C. M., Duggins, A.. J., Mahant, N., Gomes, L., Ashburner, J., McCusker, E. A. (2005) Progression of structural neuropathology in preclinical Huntington's disease: a tensor based

morphometry study. *Journal of Neurology, Neurosurgery and Psychiatry,* vol 76, 5, pp 650-655, May 1 2005.

Kremer, Berry. (2002) Clinical neurology of Huntington's disease. In Bates, G., Harper, P.S. &

Lindstrom, J.A., Bias, W.B., Schimke, R.N., *et al* (1973) Genetic linkage in Huntington's chorea. *Advan. Neurology*. 1, pp 203-208. In Bates, Harper, & Jones, (2002)

Lund, J.C. (1860) *Chorea St.Vitus dance in Saetersdalen. Report of health and medicine and medical conditions in Norway in 1860*, p.137. In Orbeck, A.L. (1959) An early description of Huntington's chorea. *Medical History*, 3: pp 165-168.

Mazumdar, P.M.H. (2002) 'Reform' eugenics and the decline of Mendelism. *Trends in Genetics,* vol 18, 1, Jan 2002. Elsevier, Netherlands. Accessed via www.sciencedirect.com Nov 2008.

Minski, L., & Guttman, E. (1938) Huntington's chorea: a study of 34 families. *Journal of Mental Science* , 48:1172-1174

Muller-Hill, B. (1988) *Murderous Science*. Oxford, University Press.

Panse, F. (1942) *Die Erbchorea; eine klinisch-genetische Studie.* Leipzig, Thieme, cited in Muller-Hill (1988) above.

Pericak-Vance, M.A., Conneally, P.M., Merritt, A.D., Roos, R., Norton, J.A.jnr, & Vance, J.M. (1978) Genetic linkage studies in Huntington's disease. *Cytogenet. Cell Genet,* 22, 640-645. In, Harper & Jones (2002), as above.

Ponder, M., Statham, H., Hallowell, N., Moon, J.A., Richards, M., & Raymond, F.L. (2008) Genetic research on rare familial disorders: consent and the blurred boundaries between clinical service and research. *Journal of Medical Ethics*, 2008:34, pp690-694. Downloaded from jme.bmj.com.on 13/10/2008

Punnett, R.C. (1908) Mendelian inheritance in man. *Proceedings of the Royal Society of Medicine,* 1: 135-168. In Bates Harper, Jones, (2002) *Huntington's disease.*

Scrimgeour, E.M., & Opitz, J.M. (1983, online 2005) Possible introduction of Huntington's chorea into Pacific islands by New England whalemen. *American Journal of Medical Genetics, vol 15*, iss 4, pp 607-613.

Solomon Andrea C., Stout Julie C., Johnson, Shannon A., Langbehn, Douglas R., Aylward Elizabeth H., Brandt Jason., Ross Christopher A., Beglinger Leigh., Hayden, Michael R., Kieburtz Karl, Kayson Elise., Julian-Baros Elaine., Duff Kevin., Guttman Mark., Nance Martha,. Oakes David., Shoulson Ira., Penziner Elizabeth., Paulsen Jane S., and the Predict-HD investigators of the Huntington Study Group. Verbal episodic memory declines prior to diagnosis in Huntington's disease. *Neuropsychologia*, *Volume 45, Issue 8*, 2007, pp 1767-1776. Downloaded from www.pubmedcentral.nih.gov/articlerender.fcgi?artid=2570315 1/3/2009

Spillane, J. & Phillips, R. (1937) Huntington's chorea in South Wales. *Quarterly Journal of Medicine,* 24 vol 6.Oxford, Clarendon Press.

Stevens, D.L. (1972) The history of Huntington's chorea. *Journal of the Royal College of Physicians,* 6: 271-282.

The Huntington's Disease Collaborative Research Group (1993). A Novel Gene Containing a Trinucleotide Repeat That is Expanded and Unstable on Huntington's Disease Chromosomes. *Cell, vol 72, 971-983, March 26 1993*, *Cell Press.*

Vessie, P.R. (1932) On the transmission of Huntington's chorea for 300 years. The Bures family group. *Journal of Nervous and Mental Disorders*, *76,* pp 553-573.

Volkers, W.S., Went, L.N., Vegter-van der vlis, M., Harper, P.S., & Caro, A. (1980) Genetic linkage studies in Huntington's chorea. *Ann. Hum. Genet.* 44, 75-79. In Bates, Harper & Jones as above.

Walker, D.A., Harper, P.S., Wells, C.E.C., Tyler, A., Davies, K., Newcombe, R..G. (1981) Huntington's Chorea in South Wales. A genetic and epidemiological study. *Clinical Genetics*, 19, pp 213-221, Munksgaard, Copenhagen.

Walker, D.A., Harper, P.S., Newcombe, R.G., Davies, K.. (1983) Huntington's chorea in South Wales: mutation, fertility, and genetic fitness. *Journal of Medical Genetics*, 20, 12-17.

THE CONSTRUCTION AND CLASSIFICATION OF 22Q11 DELETION SYNDROME

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Abstract

When Sedlackova Syndrome, Velo-Cardio Facial Syndrome, Conotruncal Anomaly Face Syndrome and DiGeorge Syndrome were initially delineated each was constructed as a separate and distinct syndrome. These classifications were defined in relation to particular patterns of physical symptoms, diagnostic criteria and clinical expertise. Advances in genetic technology over the last twenty years has led to the confirmation that all these diagnoses are caused by the same mutation, a deletion of genetic material on the long arm of chromosome 22. While these developments led to new possibilities for diagnosis, they also re-defined the boundaries between each syndrome, bringing them together under one potentially all-encompassing diagnostic label, "22q11 Deletion Syndrome". In this article I suggest that there continues to be a lack of coherency and consensus about the use of 22q11 Deletion Syndrome as a diagnostic label and explore how this label is negotiated and contested within the context of academic and medical literature, within the clinical context and from the perspective of the patient and family. This work refers to interview data collected for an ethnographic project which focused on the production and communication of genetic knowledge.

Introduction

From a social constructionist perspective, medical knowledge is understood as a production, a site of negotiation and interaction with multiple meanings. Bowker and Star (2000) highlight that medical categories and classificatory standards of disease and associated work practices are often seen as 'natural' thus rendering their construction invisible, yet these classifications have consequences. Within the medical domain, the triumvirate model of diagnosis, treatment and cure provides a dominant framework for explaining and legitimising the boundaries between health and illness. This impacts upon the patient population, but also upon those refused access to these medical categories. Explanations for the lack of bodily cause for some individuals with chronic pain for example, might focus on the imagination of the individual or the limitations in current medical knowledge and both highlight the persuasive influence of the medical model on producing the kinds of explanations that might be deemed acceptable.

Geneticization (Lippman, 1991) is a useful concept to highlight the extended reach of biomedical explanations and demonstrates the role of new technologies in the search for new biological truths. Geneticisation highlights a potential paradigm shift in biomedical knowledge with the search for cause and cure frequently couched in genetic terms, the 'gene' becoming a new, all-powerful cultural icon (Nelkin & Lindee, 1995). Within the family, the impact of the 'new genetics' (Richards, 1993) has been explored in various ways, the re-construction of kinship (Featherstone *et al.* 2006), issues of guilt and blame (Forrest *et al.* 2003) and communication strategies (Gregory *et al.* 2007). Advances in genetic technology have produced new ways of knowing disease entities. Hedgecoe (2003) and Kerr (2005) analysed the way that new knowledge about male infertility has challenged the construction of, and boundaries around 'classic' Cystic Fibrosis. However, the power of genetic technology to transform disease concepts and identities has been questioned (Bhardawaj *et al.* 2007). Developments in medical and genetic medicine are widely taken for granted as implying progress, that they will lead to improvements in diagnosis and most importantly for this paper, that they will produce one definitive explanation.

I explore the role of genetic knowledge in the construction of 22q11 Deletion Syndrome by illustrating how this 'new' label has been constructed within the medical literature as a positive move towards accuracy and inclusion. I then explore how the label might be contested and negotiated within the context of the family and in the clinic setting. Finally I discuss one

example of where genetic technologies have had a significant impact on the identity of those who might be 'ruled out' of the 'new' diagnosis.

What is 22q11 Deletion Syndrome?

22q11 Deletion Syndrome has an expansive phenotype of more than 180 potential symptoms, each of which can vary from mild to severe. The most commonly reported symptoms include congenital heart defects, mild to moderate learning disabilities, behavioural problems, cleft lip and palate, immune deficiency and distinct facial features. The label 22q11 Deletion Syndrome was introduced following the development of a technique known as fluorescent in-situ hybridization (FISH) which could identify a deletion of genetic material (a loss of many genes) on chromosome 22. Prior to this, the syndrome was diagnosed through clinical presentation and due to its expansive range of associated symptoms, various nomenclature have been developed to identify, explain and document the clinical phenotype. Thus the development of 22q11 Deletion Syndrome is frequently represented as the cumulative histories of Sedlackova Syndrome, DiGeorge Syndrome, Conotruncal Anomaly Face Syndrome and Velo-Cardio Facial Syndrome amongst others. This history is summarised briefly below.

The initial identification of the syndrome is attibruted to Sedlackova, a speech clinician who identified 26 children with hypernasal speech and cleft palate in 1955. Although at the time of publication Sedlackova's work, written in Czech, was neither widely read nor acknowledged, her role in the identification of an emerging syndrome is increasingly recognised. 'Sedlackova Syndrome' or its synonym 'Velo-Facial Hypoplasia' is sometimes offered in medical dictionaries or reference books and has remained a recognised diagnosis in parts of Europe. An association between congenital heart defects and immune deficiency was identified in 1965 by an endocrinologist, Dr Angelo DiGeorge, and this pattern of symptoms became known as 'DiGeorge Syndrome'. In 1968 Robert Strong, a paediatric cardiologist described a family with cardiac problems, learning disabilities and distinct facial features. This was an important paper because it was one of the first attempts to recognise the wide range of associated symptoms. In Japan a syndrome labelled Conotruncal Anomaly Face Syndrome (CTAF) or Takao Syndrome was identified by Dr Kinouchi and Dr Takao in 1976. The syndrome described an association between congenital heart defects and distinct facial features. In 1978 Dr Robert Shprintzen, an American speech pathologist documented a syndrome characterised by cleft palate, cardiac anomalies, learning disabilities and distinct facial appearance. The impact of this work on the

nature of the emerging syndrome was substantial, and the paper which announced the discovery of a 'new syndrome' is widely referenced and documented. In the paper, Shprintzen named the syndrome 'Velo-cardio Facial Syndrome' after the major features of the clinical phenotype (velo refers to the palate, cardio to heart defects and facial to indicate that it is associated with distinct facial features) although as is a frequent practice in the nomenclature of syndromes, the name 'Shprintzen Syndrome' is also in use.

When Sedlackova Syndrome, Velo-Cardio Facial Syndrome, Conotruncal Anomaly Face Syndrome and DiGeorge Syndrome were first delineated each was constructed as a distinct and separate syndrome. Although the similarities between the clinical presentations of each were explored to some extent, advances in genetic technology during the 1990s provided confirmation that each was caused by a deletion of genetic material in exactly the same location on chromosome 22. These developments in genetic technology have had a profound effect on the construction of the syndrome. Whereas previous classifications focused on the presentation of clinical symptoms and were represented through a range of diagnostic labels, the knowledge of shared genetic aetiology brought with it the possibility of one new, all-encompassing diagnosis.

The label CATCH22 (Wilson et al. 1993) was the first to reflect this new genetic knowledge. CATCH22 refers to the association with chromosome 22 and is an acronym representing what Wilson considered the most prominent symptoms associated with the syndrome (conotruncal heart defect, abnormal face, T-cell deficiency [immune system], clefting and hypo-calcemia). Of interest is the inclusion of immune deficiency, a symptom which might previously have been associated with the diagnosis of DiGeorge Syndrome that is absent in the label Velo-Cardio Facial Syndrome. Despite still being used in some areas of academic literature, the term has been widely rejected because of the implication that Catch22 represents a no-win situation, from the book of the same name by Joseph Heller. Burn (1999) discussed the advantages of the acronym CATCH22 including the benefits of moving away from the term 'syndrome', suggesting that this previously neutral concept had become associated with negative connotations of learning disabilities and Down's syndrome in particular. Although the label 22q11 Deletion Syndrome appears unclaimed by laboratory or clinician, since the mid 1990s, it is this label that began to be introduced into the academic and health related literature.

I have documented the emergence of 22q11 Deletion Syndrome as the lumping together of previously distinct syndromes under one diagnostic label, made possible by new knowledge of

shared genetic aetiology. However, in practice the label 22q11 Deletion Syndrome has not replaced previous explanations for the syndrome and debate continues to focus on the location of their discreet boundaries. In particular, the relationship between DiGeorge and Velo-Cardio Facial Syndrome is constructed around three types of relationships. The first is cause and consequence - DiGeorge being a secondary manifestation (a sequence) associated with the underlying causal syndrome of Velo-Cardio Facial Syndrome. The second focuses on the nature of symptoms, for example, immune deficiency is considered to be a significant factor in the diagnosis of DiGeorge Syndrome. The third, published on the Contact a Family website is to suggest that the two syndromes can be distinguished through differences in severity "from mild to moderate in Velo-Cardio Facial Syndrome, to more severe in DiGeorge Syndrome" (Habel, 2008).

One possible reason why the label 22q11 Deletion Syndrome has not been universally accepted is the continued use of the label Velo-Cardio Facial Syndrome by Dr Robert Shprintzen, a prominent clinician and prolific writer about the syndrome.

> "All of these names refer to exactly the same syndrome, and the confusion over the nosology has unfortunately resulted in clinical confusion…. VCFS is simply easier to say and write and communicate than any of the other labels and its use should therefore be encouraged" [Shprintzen, 2008]

Through his writings, Shprintzen frequently engages in what he calls 'the name' game. In the above quote, Shprintzen asserts that the diagnostic labels are interchangeable and then makes the suggestion that Velo-Cardio Facial Syndrome is the preferred term. In this way, the debate is constructed through similarity and pragmatic choice, rather than hierarchies of knowledge.

Yet while the emergence of the syndrome, and the possible relationships between the various diagnoses is well documented, there has so far been little interest in how families and health professionals approach the changing classifications. This paper offers an opportunity to explore how diagnostic labels are negotiated in practice and how they might be understood by health professionals and families. Utilising data collected from semi-structured interviews with parents and clinicians, the following section explores the impact that this reframing of syndrome boundaries has on patients and their families, and health professionals involved in diagnosis and clinical management.

How do patients and families make sense of 22q11 Deletion Syndrome?

The narrative below from one mother, highlights how the various labels of Velo-Cardio Facial Syndrome, DiGeorge Syndrome and 22q11 Deletion Syndrome might be understood. The mother who has a daughter with the diagnosis DiGeorge Syndrome explains her perspective on the relationship between the various diagnoses.

> "Well, I was – I mean my understanding is because there's VCFS as well. And my understanding is the VCFS part, it means they got the facial features which obviously she doesn't have. The DiGeorge is if they were diagnosed with the immune deficiency which she was. And then I just think that the 22q is the other bit; you know, all the – but if you don't fit into either of those two you fit into that one"
>
> [Family Interview 10]

The words of the mother are interesting from two perspectives. The first is that for this parent, the diagnosis is made meaningful in the context of physical symptoms. The range of diagnostic labels to which the mother refers are all defined in relation to clinical diagnosis and patterns of symptoms, that is, facial characteristics as a defining feature of Velo-Cardio Facial Syndrome and immune deficiency as a defining feature of DiGeorge Syndrome. In this case, the diagnoses mentioned are not considered as interchangeable 'labels', they are seen to represent distinct syndromes. The second is that '22q' is used to represent a third diagnosis rather than an 'umbrella'. I suggest that this perspective might be understood through recognising that 22q is constructed through genetic knowledge rather than clinical knowledge. The narrative above might indicate a deficit in the language available to the mother to describe what 22q means, reducing the ability to understand where 22q 'fits' with the other diagnostic labels.

The following account shows how parents of children with the syndrome might take a different approach to the diagnosis. In the interview extract below, the mother and father of a child diagnosed with DiGeorge syndrome discuss their perspective on syndrome nomenclature. The parents indicate their understanding that the diagnoses of DiGeorge Syndrome, Velo-Cardio Facial Syndrome and '22q11' might be interchangeable labels. Indeed, the father explains that it his experience that different consultants might use different terms, where one might use DiGeorge Syndrome and the other might use 22q11. While this family express their competence

with the different diagnoses, they also highlight the decision making processes that occur when selecting which label to use.

> Father: I think – I think we just call it DiGeorge. I think it makes it easier to explain like, you know, just to your friends, family rather than…
> Mother: Yeah, 22q11, yeah or velocardiofacial syndrome is bit of a mouthful as well.
> Father: Yeah, I think we just, you know, choose to say that 'cos any of his - in fact they vary I'm sure from different consultants. Some of them say DiGeorge and others say 22q and my understanding was it was all the same thing.
>
> [Family Interview 21]

In the above narrative, the family appear to suggest that there is a range of diagnostic labels at their disposal. Although the words of the parents indicate that these diagnostic labels might be understood as "the same thing", the father indicates that the label of DiGeorge Syndrome proves more useful in terms of communicating the diagnosis to friends and family, and throughout the interview the family continued to use the one label of DiGeorge Syndrome. The consistency in the relationship between parent and diagnosis is apparent throughout the parental interviews conducted for this project where, in practice, or at least in their talk, they continue to use the one label that offers the most significance. I suggest that where genetic knowledge has had a significant impact on the construction of the syndrome classification, parents continue to express their understandings of the diagnostic label in terms that are meaningful to them, that is, within their everyday experience of physical symptoms and social relationships.

How is the diagnostic 22q11 negotiated within the clinical context?

While the families might express their understanding of 22q11 Deletion Syndrome in terms of everyday, grounded experience the next section explores how the diagnostic label might be negotiated within the clinic context. The extract below demonstrates the approach taken by one clinician who works at a specialist clinic for patients with the syndrome.

> Interviewer: Does it actually matter to the parents that there is this 22q11 yet they might have had a diagnosis of Di-George?
> Respondent: Well in that instance it is better to use Di-George because they have received the diagnosis and that is how they will identify themselves, that is how they will

> orientate but I think that to make sure there is no confusion it is important to explain the terminology. You know what is 22q11, what is the difference between that and Di-George, is there a difference? It is probably something that we tend to forget, the language that the families use when they are talking about the diagnosis and our tendency…I am not sure what [colleague] says actually but I think that it is important that we all use the same language when we are talking to the families, I am not sure that we do that.
>
> [Interview with Dr Jones, Psychologist]

Dr Jones demonstrates how clinicians might actively engage in the management of diagnostic labels. Rather than focus on diagnostic criteria, the clinician talks about the language of diagnosis. Dr Jones goes beyond constructing the diagnosis as a 'label', referring to the significance of a particular diagnosis for the patient and how it might become an integral part of their identity. However, the words of Dr Jones highlight that the lack of consensus might cause tension between respecting the patient/parent choice of diagnostic, yet ensuring that all clinicians within the clinic "use the same language". Where developments in genetic technology impact on syndrome construction and nomenclature, it is clear that the clinic will continue as an important site of negotiation between the clinician's and patient's understandings and meanings associated with diagnosis.

I have thus far considered the ways in which changing nomenclature associated with the syndrome might be understood by families and how this might be negotiated within the clinic. I now consider the potential impact that re-constructing diagnostic classifications have on the identities and relationships of individuals and their family members. Novas and Rose (2000) provide an instructive account of the transformative effects of genetic technology including the modification and shaping of patient identities. Finkler (2000) used the concept of 'patients without symptoms' to show how carriers of a genetic illness, or those who might be 'at risk' of developing an illness are constantly being re-defined through medical categories. Miller (2005: 2543) highlights that for some conditions such as tuberous sclerosis "genetic mutations are a sufficient but not a necessary condition for entry into the disease category" whereas for others, such as Klinefelter, Fragile X and Turner Syndrome the presence of the genetic mutation alone becomes the diagnostic criteria (Whitmarsh, 2007). I suggest that 22q11 Deletion Syndrome might be included in the latter category, where diagnostic decisions are based on whether the

patient has the deletion as determined through the FISH test, irrespective of the symptoms that they present, and this I suggest, has implications for the identity of the patient population.

Whereas 22q11 Deletion Syndrome might be identified as an inclusive and expansive term incorporating previous diagnostic categories, the narrow focus on genetic aetiology, the re-construction of the syndrome around genetic diagnosis, has proved potentially problematic for diagnostic clinicians and patients. One example of this is the estimation that up to fifteen percent of the population with the clinical phenotype of 22q11 Deletion Syndrome do not have the specific deletion (Hay, 2007:138). Suggestions as to why this might be the case include the possibility that an individual might have a smaller deletion which can not yet be identified through current technology, a deletion in a different location or on a different chromosome, or the individual might have a different syndrome entirely.

Within the clinical context, the return of a negative FISH result can create diagnostic uncertainty. The American Heart Association advise that where a genetic cause has not been identified, diagnosis should be on the basis of clinical presentation, in which case, it might be assumed that the diagnosis of DiGeorge or Velo-Cardio Facial Syndrome becomes appropriate. Whereas returning to a diagnosis of DiGeorge or Velo-Cardio Facial Syndrome is one way of making sense of the symptoms, for some, this step would render a diagnosis meaningless. For example, a clinician to whom I spoke indicated that in the absence of the deletion, the patient would not be given any diagnosis at all.

> "If they haven't got the appropriate micro deletion, I mean they're likely to have something else aren't they. That is why I think it is not particularly helpful, if somebody hasn't got a particular condition and then [to] say that it is 'something like', how is that benefiting them anyway? Because you can't tell what the risks are, you can't tell what the side effects are, you can't tell them what the complications are because that child is probably never going to get them."
>
> [Interview with Dr Evans, Geneticist]

Dr Evans suggested that the negative result of the genetic test is taken as evidence that the clinical symptoms are related to a different syndrome entirely and that the diagnosis of 22q11 Deletion Syndrome would be misleading. However, as can be seen through the extract below, not all clinicians take this approach. Whereas Dr Evans focused on the primacy of the genetic

diagnosis, the clinician below talks about the possibilities for inclusivity associated with a clinical diagnosis.

> "I guess the difficulty that I had with calling it 22q11 Deletion Syndrome is that not everybody has a deletion. And that is in a sense why I don't like the term, I feel that VCFS is a clinical diagnosis which includes all those people who have a deletion but also those who don't."
>
> [Interview with Dr Rees, Psychiatrist]

The extract above demonstrates one way in which uncertainty might be negotiated, and that for this health professional, the provision of a clinical diagnosis (Velo-Cardio Facial Syndrome) in the presence of a negative genetic result would continue to prove useful. The results of a genetic test might exclude a certain percentage of the patient population from accessing the label '22q11 Deletion Syndrome', yet access to the syndrome diagnosis is still possible if the diagnosis of DiGeorge or Velo-Cardio Facial Syndrome can be negotiated. These two quotes together reveal a tension between clinical and genetic diagnosis.

But what is the appropriate stance for a clinician to take? In the extract above, Dr Evans suggests that the provision of a diagnosis of 22q11 Deletion Syndrome in the presence of a negative FISH result would not provide the family with useful information about the diagnosis or prognosis. The emphasis for Dr Evans is genetic diagnosis, and genetic accuracy. Dr Rees however takes a pragmatic approach, focusing on clinical application. For the patient, the exclusion from a disease category can be significant. Although a diagnostic label can have a negative impact such as stigma or exclusion (Gillman, 2000) a diagnosis can be desirable in terms of accessing resources, making sense of an illness and future planning. For those without the deletion who are 'ruled out' from the diagnosis of 22q11, the suggestion that they may have another as yet unknown syndrome can have serious implications, it may affect an individual's 'imagined future' or create uncertainty about reproductive risk (Kobrynski, 2007). The debate surrounding diagnostic possibilities for the syndrome, as revealed when there is tension between a clinical diagnosis and genetic diagnosis, demonstrate how scientific and medical knowledge is performed through local negotiation (Knorr-Cetina, 1983).

The Impact of Genetic Technology

The concept of geneticisation has been explored as a shift from the 'clinical gaze' to the 'genetic gaze' (Atkinson & Gregory, 2008) reflecting a perceived relocation of the diagnostic process from clinic to genetic laboratory and a shift in diagnostic expertise from the clinician to the laboratory technician. However ethnographies of clinical practice have demonstrated that this is not necessarily the case, and this article builds on these observations. The clinic continues to play a significant role in the construction of medical classifications (Latimer et al. 2006) and in the diagnostic process (Featherstone et al. 2005).

Within the academic and medical literature, the first instalment in the history of 22q11 Deletion Syndrome is represented as being closely bound to the professional or personal interests of each clinician and researcher. A frequently used analogy to illustrate the multiple symptoms and potential diagnoses is where a blind man ventures across an elephant. The elephant can be characterised by big ears, long trunk, hard tusks and rough skin, depending on which part is touched first or considered the most significant. This image is used to show how "each person was accurate in describing his/her own area of interest, but none was able to see the big picture" (McDonald-McGinn & Zackai, 2005: 201).

The image that is portrayed provides an interesting explanation for the way in which 22q11 Deletion Syndrome has been constructed. The image reflects on the role of a range of clinical experts involved in mapping out their own experience of the syndrome and suggests that these separate diagnoses were pieces of a jigsaw puzzle, which on their own did not make up the whole picture. Significantly, it might be inferred that this 'big picture' which has thus far eluded the scientist and clinician, can only be known through genetic expertise.

There is no doubt that advances in genetic technology has had a significant impact on the construction of 22q11 Deletion Syndrome. The concept of 'lumping and splitting' (McKusick, 1969) is a useful tool for de-constructing 22q11 Deletion Syndrome as a label which envelopes previously distinct multiple diagnoses. The emphasis on progress, a potential move from clinical to genetic explanations might suggest the emerging supremacy of the label 22q11 Deletion Syndrome. Yet there is no national nor international consensus about which term is preferable, and I have demonstrated that rather than leading to the redundancy of previous diagnoses, in practice and in the literature, the label 22q11 Deletion Syndrome has simply added to the range

of diagnoses available to the clinician and family. The diagnostic label of 22q11 Deletion Syndrome is negotiated, by the patient within the context of grounded experience, and by the clinician within the context of professional expertise and judgement. The decision making process has been highlighted through the potential tension of a mismatch between phenotype and genotype.

This article suggests that genetic technology has re-framed 22q11 Deletion Syndrome through the construction and reconstruction of boundaries. With the range of specialisms involved in the management of the syndrome, reflecting a wide variety of experience and expertise, it is perhaps not surprising that there are differences in the approach to the syndrome, its diagnosis and management. The new genetics has produced techniques which have added to the diagnostic tool kit for the clinician, has the potential to produce a non-symptomatic 'patient' while at the same time questioning the legitimacy of some patients' claim to the diagnosis. However, the meanings associated with 22q11 Deletion Syndrome can not be understood through a reference to genetic technology alone and the expectation that genetics might provide definitive and certain knowledge is in sharp contrast to the reality of diagnostic negotiations. I suggest that genetic knowledge has only partially re-constructed the syndrome and that a dual system of categorisation exists in practice – one that is based on the knowledge of shared genetic aetiology, and one that remains firmly fixed in the context of physical symptoms.

Bibliography

American Heart Assocation [www] http://www.americanheart.org/ presenter.jhtml?identifier= 3018193 [accessed 11/12/2007]

Atkinson, P. and Gregory, M. 2008. Constructions of Medical Knowledge. In: Holstein, J. & Gubrium, J. eds. *Handbook of Constructionist Research*. Guilford Press. pp.593-608

Bharadwaj, A., Atkinson, P. and Clarke, A. 2007. Medical classification and the experience of genetic haemochromatosis. In: Atkinson, P. et al. eds. *New Genetics, New Identities*. Oxon: Routledge. pp.120-138

Bowker, G. C. and Star, S. L. 2000. *Sorting things out: Classification and its consequences*. MIT.

Burn, J. 1999. Closing time for CATCH22. *Journal of Medical Genetics.* 36: pp.737-738.

Featherstone, K., Latimer, J., Atkinson, P., Pilz, D. and Clarke, A. (2005) Dysmorphology and the spectacle of the clinic *Sociology of Health and Illness.* 27 (5) pp.551-574

Featherstone, K., Atkinson, P. A., Bharadwaj, A. and Clarke, A. J. 2006. *Risky Relations: Family, Kinship and the New Genetics.* Oxford: Berg.

Finkler, K. 2000. *Experiencing the New Genetics.* University of Pennsylvania Press.

Forrest, K. Simpson S. A., Wilson, B. J., Van Teijlingen, E. R., Mckee, L., Haites, N. and Matthews E.2003. To tell or not to tell: barriers and facilitators in family communication about genetic risk. Clinical Genetics. 64: pp.317-326.

Gillman, M., Heyman, B. and Swain, J. 2000. What's in a name? The Implications of Diagnosis for People with Learning Difficulties and their Family Carers. *Disability & Society* 15(3): pp.389-409.

Gregory, M., Boddington, P., Dimond, R., Atkinson, P., Clarke, A. and Collins, P. 2007. Communicating about haemophilia within the family: the importance of context and of experience. *Haemophilia.* 13 (2): pp.189-198.

Habel, A. 2008. 22q11 Deletion Syndromes. [www] http://www.cafamily.org.uk/ Direct/ 22.html [accessed 10/10/2007]

Hay, B. N. 2007. Deletion 22q11: Spectrum of Associated Disorders. *Seminars in Pediatric Neurology* 14: pp.136-139.

Hedgecoe, A. M. 2003. Expansion and uncertainty: cystic fibrosis, classification and genetics. *Sociology of Health & Illness.* 25(1): pp.50-70.

Kerr, A. 2005. Understanding genetic disease in a socio-historical context: a case study of cystic fibrosis. *Sociology of Health & Illness* 27(7): pp.873-896.

Knor-Cetina, K. D. 1983. The Ethnographic Study of Scientific Work: Towards a Constructivist Interpretation of Science. In: Knor-Cetina, K.D. and Mulkay, M. eds. *Science Observed.* London: Sage Publications. pp.115-140.

Kobrynski, L. J. and Sullivan, K. E. 2007. Velocardiofacial syndrome, DiGeorge syndrome: the chromosome 22q11.2 deletion syndromes. *The Lancet* 370: pp. 1443-1452.

Latimer, J., Featherstone, K., Atkinson, P. A.,. Clarke, A, Pilz, D. and Shaw A. 2006. Rebirthing the clinic - The interaction of clinical judgment and genetic technology in the production of medical science. *Science Technology & Human Values* 31(5): pp.599-630.

Lippman, A. 1991. Prenatal Genetic Testing and Screening: Constructing Needs and Reinforcing Inequities. *American Journal of Law & Medicine* 17(1/2): pp.15-50.

McDonald-McGinn, D. M. and Zackai, E. H. 2005. Genetic counselling. In: Murphy, K.C. and Scambler, P.J. eds. *Velo-Cardio-Facial Syndrome: A Model for Understanding Microdeletion Disorders.* Cambridge University Press.

McKusick, V. 1969. On lumpers and splitters, or the nosology of genetic disease. *Perspectives in Biological Medicin.e* 12(2): pp.298-312.

Miller, F.A., Ahern, C., Ogilvie, J., Giacomini, M. and Schwartz, L. 2005. Ruling in and ruling out: Implications of molecular genetic diagnoses for disease classification. *Social Science & Medicine.* 61: pp.2536-2545.

Nelkin, D. and Lindee, M. S. 1995. *The DNA Mistique.* New York: W.H. Freeman & Co.

Novas, C. and Rose, N. 2000. Genetic risk and the birth of the somatic individual. *Economy and Society.* 29(4): pp.485-513.

Richards, M. P. M. 1993. The new genetics: some issues for social scientists. *Sociology of Health & Illness* 15: pp.567-587.

Shprintzen, R. J. 1978. A new syndrome involving cleft palate, cardiac anomalies, typical facies, and learning disabilities: velo-cardio-facial syndrome. *Cleft Palate Journal.* 15(1): pp.56-62.

Shprintzen, R. J. 2008. Velo-Cardio-Facial Syndrome: 30 Years of Study. *Developmental Disabilities Research Reviews*. 14: pp.3-10

Whitmarsh, I., Davis, A.M., Skinner, D. and Bailey, D.B. 2007. A place for genetic uncertainty: parents valuing an unknown in the meaning of disease. *Social Science & Medicine.* 65(6): pp.1082-1093.

Wilson, D., Burn, J., Scambler, P. and Goodship, J. 1993. DiGeorge syndrome: part of CATCH 22. *Journal of Medical Genetics.* 30(10): pp.852-856.