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Dysmorphology and the Spectacle of the Clinic

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Dysmorphology and the Spectacle of the Clinic

Abstract

Dysmorphology is the medical study of abnormal forms in the human and is concerned with the identification and classification of a variety of congenital malformations. Such diagnostic work rests on the inspection of images of affected individuals. Based on physical appearance individuals are classified in terms of a wide range of conditions, often with 'exotic' nomenclatures. This paper will describe the features of clinical dysmorphology and the process of classification. It derives from an ethnographic study of clinical consultations and meetings among medical geneticists in UK hospitals. We suggest that contemporary dysmorphology can be understood in terms of long-standing forms of medical knowledge, medical representations and medical discourse. Notwithstanding the new forms of technology provided by genetic science, 'the clinic' still asserts its symbolic and functional power: the 'gaze' of the clinician and the clinician's warrant of personal knowledge exert their influence. The adjudication of dysmorphology is a contemporary exemplar of the spectacular.

Dysmorphology and the Spectacle of the Clinic

Dysmorphology

Contemporary dysmorphology preserves many features of earlier forms of physiognomy and iconography; however, in recent years, it has also been subject to increasing technical change. It thus stands at the crossroads of the old clinic and the new technologies of genomic science. Dysmorphology represents a traditional clinical area that is increasingly using genetic technologies in ways that redefine clinical work. Diagnosis and clinical classification are being reshaped by genetic technologies. Thus, clinical dysmorphology parallels other clinical areas such as neurology, oncology and infertility and represents an important site where clinical medicine and genetic science intersect.

Dysmorphology refers to the professional discipline of delineating disorders affecting the physical development of the individual, before or after birth, and includes the recognition of specific patterns of physical features in patients with a range of problems (Aase 1990). These features may be associated with abnormalities but need not be abnormal in themselves. However, particular patterns of physical features have come to be associated with underlying systems abnormalities such as heart defects, or delayed intellectual development. Patients are mainly babies, children and teenagers or young adults. When patterns of malformations are deemed to have reached a level of regularity across different cases and are thought to arise from a single underlying pathogenetic mechanism, they are named as a syndrome. There are several thousand named syndromes currently held within international clinical databases and textbooks. The majority of syndromes are associated with a genetic basis.

The specific features that characterise clinical dysmorphology include: the recognition and classification of specific patterns of facial and other physical features; ongoing classification based on clinical diagnosis and examination; increasing use and interpretation of molecular tests in diagnosis and clinical classification; decision-making and assessment distributed and networked between different experts (including scientists and clinicians) at local, regional and national levels; and variable outcomes, for example, the introduction of new clinical categories and diagnostic labels. Thus, the examination of the field of dysmorphology reveals a speciality that displays the interaction of genetic technologies and clinical judgement. Shaw's analysis of the exercise of judgment by dysmorphology specialists provides a parallel example to our own (Shaw 2003).

In the course of this paper, we shall locate the visual culture of dysmorphology within the wider history of medical representation. We shall consider this in terms of the 'spectacular display' of the clinic. We shall discuss the dysmorphology experts competence is 'seeing' cases and interpreting visual representations. This is in turn repeated in the expert's right to make 'oracular pronouncements' concerning the patient's characteristic appearance and its clinical significance. We go on to discuss the intersection of genetic technologies and clinical judgement in the identification of dysmorphic conditions. We conclude by affirming the continued significance of 'the clinic' and the importance of resisting premature or over-simplified appeals to geneticization or technologically driven reductionism.

The spectacle of the clinic

For centuries, the clinic has been a site for the spectacular display and representation of bodies, organs and pathologies. The clinical spectacle has taken many forms and these include the public dissection and the anatomy lesson; the clinical lecture; the

ward round; the teaching round; the grand round and the clinico-pathological conference. Michel Foucault (1982) wrote vividly on the clinical 'gaze' (*le regard*) in the development of the modern clinic. He suggested that during the rise of the distinctively modern university hospital in post-revolutionary Paris, the patient's bedside became a site of privileged perception. The development of technologies of inspection coupled with the inception of clinical pathology meant that classical nosographies of medicine in the *ancien régime* became supplanted by a new clinical medicine. From this point, disease became situated within specific organs; diseases and their course could be correlated precisely with pathological findings; and 'the lesson of the hospitals' created a radically new mode of medical perception. As King (1982) and others have suggested, the distinctive modes of perception and inference of the clinic are by no means exclusive to the particular moments and transitions identified by Foucault himself. They remain deeply embedded in the culture and practice of medicine.

Consequently, we need to preserve Foucault's concern with the technologies of inspection and medical inference, while continuing to investigate the modalities of medical perception (cf. Casper and Berg 1995; Berg and Mol 1997). We concentrate on the spectacular presentation and representation of patients, their bodies and their identities within the clinical space defined by contemporary genetic medicine. We suggest that a broad historical and cultural pattern can be traced that brings together the *spectacular display* and the *oracular pronouncement* as long-standing (although by no means immutable) features of medical knowledge and the importance of a deeply entrenched visual and oral culture in the creation and transmission of medical knowledge. We trace the intersection of visual culture and nosographic classification in the genetic clinic.

The social *forms* of the spectacle are various, and have their own *longue durée*. The early modern anatomy lesson (Sawday 1995; Richardson 1988) is a classic case in point: the anatomy theatres of Padua, Leiden and elsewhere are physical embodiments of spectacular history and dissections themselves were 'staged events, exuding an exciting aura of wonder and morbid fascination' (Kemp and Wallace 2000: 23). The recent resurgence of interest in the anatomical imagination and the relationships between art and anatomy has reaffirmed the cultural significance of the spectacular display of the body itself and its representations. The genealogies of representation run from fine-art anatomical drawings, to the engraved plates of anatomical atlases, to wax anatomical figures, through to modern imaging technologies (see e.g. Elkins 1999; Kemp and Wallace 2000; Hamilton and Hargreaves 2001). The new genomics has generated further convergence between the aesthetic and the scientific (Anker and Nelkin 2004).

There is a long tradition in which 'patients' are translated into reproductions and representations. The medium of photography has provided a rich vein of spectacular representations of individual patients and their characteristics. There have been, of course, many photographic representations of organs and lesions, used to illustrate textbooks and atlases of pathology. The type case and the classic presentation have been captured through photography from the earliest years of photographic technology and this technology has been used to compile extensive typologies of characters and social types. Photography provided a mechanical means that complemented and then supplanted fine-art traditions in the representation of physiognomy.

The practice of physiognomy has a long history. The identification of character and temperament through physical appearance has been deeply rooted in the iconography of Western art and science. Leroi (2003) provides a recent guide to the

long history of representations of abnormal appearances and the particular fascination they have held for medical science and the popular imagination. Appearance has long been thought to reveal the inner character of the person; as Kemp and Wallace (2000: 94) suggest; '... philosophy, science and medicine have been consistently mobilized over the ages to provide a framework of explanation of how inner is expressed in outer'. Photography paralleled and expanded upon the representational practices of the fine arts by depicting types, characters and pathologies. The image of the racially inferior specimen, the sexual stereotype or the delusional inmate became fixed on the photographic plate.

The modern clinic is now suffused with images of patients as well as images and representations of their tissues, organs and lesions. The range of technologies has been expanded and the visual penetration of the 'inner' and the 'microscopic' has been extended. The body is variously sectioned, imaged, stained, visually enhanced by false colour, and rendered visible through a diverse range of technologies. However, the photographic image of the individual patient, and the inspection of her or his appearance persists. Such presentations are also sites for the enactment of *oracular authority* by genetic scientists and clinicians (cf. Atkinson 1995, 2004).

Within this paper, we show how visual display and the rhetoric of clinical authority are long standing features of dysmorphology to demonstrate that even when new molecular technologies are used, clinical judgement is still central. The material drawn on in this paper is taken from a wider ethnography of clinical genetic work and is based on fieldwork carried out within a clinical genetics service based in a well-established UK regional genetics service.

Methods

Within this ethnographic study, one clinical genetics team and their patient population were followed over a period of 9 months, from November 2002 to July 2003. Clinic consultations (n=37) were observed within clinics (n=12) based in three local hospitals. Although the caseload of the clinical team was not dedicated to dysmorphology cases, a large number of their referrals (32) involved dysmorphology. The average length of time allocated to each consultation was one hour and this generated 44 hours of observation, yielding notes that included near-verbatim text. We also observed local professional dysmorphology meetings (n= 6) where cases were presented and discussed. In addition, a large number of less formal encounters between professionals was observed.

Selection criteria included referral for dysmorphic features, willingness to participate, and the clinical team's assessment that participation was appropriate. Because of the nature of the conditions under observation, the majority of patients were children, which necessitated full involvement and consideration of parents and other family members in the study. Where feasible, informed consent was obtained from all family members present in the clinic, with the exception of a number of occasions where very young children were involved or where the child's learning disabilities meant that it would have been unrealistic or unduly invasive to seek consent. In such cases, proxy consent was obtained from parents. This project was approved by the Multi-centre Research Ethics Committee. All names have been changed to preserve anonymity.

Spectacular display

As we have emphasised, visual display is a long-standing feature of medical knowledge. In this section, we describe the role of visual representation in the creation and transmission of medical knowledge. Within clinical genetics, the

photographic image of the individual patient, and the inspection of her or his appearance persists. It can be found in the presentation and discussion of the dysmorphic patient and the adjudication of dysmorphological nosography.

Dysmorphology has traditionally classified pathologies on clinical grounds but has increasingly made use of genetic technologies. However, molecular genetic tests do not necessarily enter the clinical process until after the team have reviewed other materials and sometimes they do not enter at all.

The most prominent visual technology utilised in the process of adjudication and classification is the photograph. There are two types of photograph employed by the clinical team, slides taken during the clinical consultation and family photographs collected by the Genetic Nurse Specialist at the initial home visit. The photographs together with the letter of referral and the report of the home visit are commonly used to initiate the process of classification *before* the individual attends the clinic for consultation. As one of the consultants put it during a local dysmorphology meeting '*I regard it [the camera] as my stethoscope*', adding that she and many other colleagues had purchased their own cameras. [Meeting 5]

Where photographic evidence is available (either accidentally within group family photographs or because they are specifically requested by the team), the physical features of other family members are examined, discussed and compared. This is part of the process of establishing whether a feature is within the normal range, the team 'read' the photographs for visual signs of dysmorphism.

The clinical consultation concerns a five year old boy who has been referred with '*a large head and learning difficulties*'. After taking a history, the consultant examines him; looking closely at his ears, his forehead, his eyes, using a small light to look in his mouth to examine his teeth and tongue. She examines his limbs, compares his fingers, and toes and after noting his '*loose*

joints,' [hyperelastic joints may be an indication of an underlying syndrome] she measures his head circumference with a tape measure. She inspects and notes his '*thick hair*' and '*hairy back*' [we can see a fine down of dark hair running down his spine] and asks the boy's mother about other family members- the father is '*hairy*' and his sister has similar hair. She compares previous slides taken of the boy with a photograph the mother gives her of his sister, adding '*we have to disentangle what is familial, he looks like his sister and his dad is hairy. There's not much to say about his face, apart from his straight eyebrows*' [an unusual eyebrow configuration can be an indication of a number of syndromes], *but it could just be him*'. [Clinic 1, patient 3]

In this case, the clinician is looking for ways to distinguish physical features, what is normal and what is abnormal. This assessment is based on the clinician's experience of 'seeing' such features and being able to distinguish when a feature deviates from the normal range. However, such features are also compared with the child's immediate family - it may be outside the normal range, but a benign feature within this particular kindred. In the case above, the child's hair distribution is evaluated and the unusual hairiness on his back are commented on, as is the general density of his hair. It is important to establish whether such a feature is familial because if not, it may be a clue to an underlying syndrome. For example, very sparse hair is associated with a number of syndromes that involve teeth and other ectodermal structures. However, the clinician is cautious; the child may just have hair like his father and sister. This does not mean that she brings to an end the search for an underlying syndrome, rather she moves on to another potential classification, the 'straight eyebrows' may be significant.

Slides are taken during the clinical consultation and usually feature the front of the face, pictures of each profile and close-ups of feet, hands and any other

interesting features the clinician feels may be significant such as toes, fingers or eyebrows. These slides are filed within the patient's medical records and can be held within each clinician's own collection of syndromes and features. They are a visual record of the child and are used both to initiate the process of classification and as an ongoing record of their development and changing features. They are routinely taken to be scrutinised and interrogated for 'clues' elsewhere, most commonly, the local dysmorphology meeting (cf. Shaw, Latimer, Atkinson and Featherstone 2003):

At the local dysmorphology meeting two consultants and three juniors are present in the room. The lights are turned off and the projector switched on. We all move our chairs to face one wall where a fuzzy out-of-focus picture of a face is projected. A junior is at the controls, however; she cannot get the picture to focus and the two consultants help. Finally, it comes into focus and shows a head and shoulders of an attractive little boy, smiling into the camera. They team exclaim variously that he is 'a *cutie*' and '*lovely*'. The first consultant gives a history '*premature baby, 14 months, doing well*'. The next slide shows a close up of one hand showing four fingers, two fused together. The consultant describes this as '*syndactyly of the right hand*' [Syndactyly is the fusion of one or more digits into a single mass] '*small mouth, small ears, broad first toes.*' She adds that she '*thought he might have some boney duplication underneath*' she is considering discharging the patient because the problems are '*unlikely to reoccur*' in a future pregnancy and asks the second consultant for her opinion. They decide to x-ray the hands and keep the child within the clinic. [Meeting 5, case 1]

In the extract above, the consultant has identified a number of subtle signs, however, although these features generally suggest an underlying genetic change, they do not

point to a specific syndrome. In such cases, the consultant routinely confers with colleagues locally. An important feature of these slides is that other professionals can see them; patients are presented *in absentia* and classification can be based on visual and laboratory evidence alone. They are a form of representation of the patient that can travel to other specialists and can be presented to colleagues locally, nationally and internationally if a case proves particularly subtle, interesting or difficult to classify.

Visual representation is fundamental to the creation and transmission of medical knowledge (Cartwright 1995; Kevles 1997). As we have shown, the recognition and adjudication of pathology in professional encounters are collective, there is a division of labour among different medical specialties, and there is a hierarchical division of labour among the medical practitioners: juniors 'present' and seniors adjudicate. These slides are also collected for teaching purposes; the consultants use them to transmit the process of adjudication and classification to juniors in demonstrating how to '*see*' syndromes or features.

An important part of the process of developing expertise is the collection of types and cases. The collection of photographs and slides is an important part of the ongoing classification of syndromes. These genetic syndromes are in the making; medical knowledge is partial and produced within the clinic.

At the local dysmorphology meeting, a slide of a '*very pretty*' little girl is presented by one of the consultants. She reports the case history and then adds that '*she's got a known diagnosis*'. The trainees comment broadly on her facial features but after further prompting by the consultant they fail to suggest any investigations they would carry out. The consultant states that she has '*an 8p terminal deletion*', adding '*we're going to have to start collecting them and putting them together*'. [Meeting 6, case 7]

Here the consultant coaches the juniors to observe, transmitting her knowledge of what to look for and how to ‘see’ features. The collection of cases such as this is part of the ongoing process of the assembly of both individual cases and the delineation of syndromes. Categories are refined and the ‘looks’ within syndromes are reassessed by considering them in the light of new technologies and other diagnostic information identified from collected cases.

It is important not only to ‘see’ a syndrome but also to have collected a case, that is, to make a diagnosis and to have a photographic record. The images become part of the dysmorphologist’s personal collection and if the syndrome is particularly ‘rare’ or interesting, the case can be presented at national or international meetings.

As soon as we gather for the local dysmorphology meeting, one of the consultants cannot contain herself she is so excited, she exclaims *‘I’m bursting to show these slides...I’ve got one I’m desperate to show’*. The trainee moves the slides on to show a picture of an 8-year-old girl with short brown hair smiling for the camera. The consultant asks the trainees *‘anyone like to make a diagnosis?’*, when they remain silent she runs through the case history. The slides move on to show a side head shot and a close up of her hands palm down. The trainees’ comment vaguely on her hands and nails following a slide of the girl’s left foot and a close up of her toes. The consultant gives them a few more hints and then shows them a slide of the girl’s mother [a slide showing the mother’s profile] and comments on her *‘striking’* eyebrows that have a high and prominent arch. There is silence until the other consultant breaks the spell *‘we’re talking about Kabuki aren’t we’* [Kabuki syndrome was first reported in 1981 and since then over a hundred cases have been identified. It is so called because the facial features of the

individuals resemble the make-up of Japanese Kabuki actors. These features include long palpebral fissures of the eyes (a narrowing of the space between the upper and lower eyelids), arched ‘interrupted’ eyebrows and prominent eyelashes, giving the impression that the patient is wearing eyeliner. Problems associated with the syndrome are growth deficiency, mild learning problems and cardiac defects]. They agree that the mother and child are affected, that this is ‘*a good family*’, ‘*mum’s got the full house*’, and because there are very few reports in the literature of this ‘rare’ syndrome being transferred from parent to child, it would be an ideal case to write up. [Meeting 4, case 4]

This case is particularly interesting to the clinical team for a number of reasons. To have identified a rare or unusual syndrome such as this has the distinction of increasing expertise generally, but actually ‘seeing’ this syndrome also confers a specific authority over the syndrome. In this case, the mother is a ‘classic’, she had ‘the full house’; that is, she displays all the main features and abnormalities associated with the condition. However, because the daughter is similarly affected this case has the added distinction of also being ‘rare’ because the known aetiology of the syndrome is that it occurs sporadically and is not familial.

Seeing a number of such rare cases and securing that expertise through presentations and publications can lead to the local, national or international recognition of a clinician’s skill to adjudicate upon difficult diagnostic decisions such as cases on the boundaries of a syndrome. Syndromes are also named by or after a clinician (for example Down’s syndrome) and the clinicians themselves can be named after a syndrome. The team occasionally discussed whether to send borderline cases to such experts as the final arbiter of a diagnosis. For example, the ‘White Matter Queen’ (an expert at interpreting brain anomalies), the ‘Rett Queen’ (an expert on Rett syndrome, a common cause of profound intellectual disability in girls. It usually

presents with stagnation of development in infancy and then a progressive loss of skills leading to profound mental and physical disability) and the ‘Angelman Queen’ (an expert on Angelman syndrome, a condition which causes severe developmental delay, and is characterised by an abnormal gait, characteristic facial features and often inappropriate laughter). Similarly, colleagues may have a local reputation for ‘seeing’ particular syndromes. For example, during a case review the consultant suggests that they send photographs of the child to a colleague who ‘*is good at spotting Marfan’s*’ (the physical features of Marfan syndrome include a tendency towards tall stature, joint laxity, high palate and dental crowding, and long fingers and toes). The opinion of such colleagues is treated with a greater degree of trust, they were often asked to adjudicate on borderline or disputed cases and such classifications are then less likely to be called into question.

The photograph is also a portable form of representation of the individual, which can be presented, mailed or emailed to other experts, locally, nationally or internationally. Discussion and adjudication of cases can be based on this technology alone. Even if the consultant cannot ‘see’ a syndrome, one feature may be enough to indicate that the problems have a genetic base and should be investigated further. Thus, images of the individual or their various parts such as hands or feet can be shown to other experts.

A little boy with severe developmental delay [Sebastian] is attending the clinic with his parents. After taking a case history and carrying out a physical examination of the boy the consultant tells the parents that she ‘*hasn’t seen anything distinct with Sebastian to say he has a particular syndrome, all the clues we look for he doesn’t have a pattern*’. The consultant decides to take more photographs and takes two frontal photographs of his face, the left side of his face and a close up of his hands because she notes he has an ‘*interesting*

thumb'. She adds that when the pictures are developed she will '*show them to the others and then work out what would be appropriate*' for the next stage.

After the consultation the team review the case, the consultant adds '*we've got to do something with that one*'. [Clinic 5, patient 1]

A genetic diagnosis is not dismissed; rather the combination of a family actively seeking a diagnosis and one feature (in this case an 'interesting thumb') is enough to seek the advice of other experts. The case is presented at the local dysmorphology group meeting and the consultant gives the history and shows slides of the boy. The group have no other suggestions that could link this feature with a diagnosis, but they decide to recommend the use of another visual technology- an MRI scan of his brain.

Although there are informal occasions where cases are discussed and adjudicated upon - in the corridor, in the car on the way to a clinic and over lunch - the local dysmorphology meeting is an important site for the transfer of knowledge. Typically, cases are presented for the purposes of teaching juniors in two ways: the consultants present the slides of one of their patients and juniors are asked to construct the case or juniors present and work up the case and the consultants adjudicate.

These monthly meetings take place with everyone sitting in a semi-circle facing the screen. The lights are dimmed, the projector emits a loud clunk and the first photograph of a child is illuminated onto the screen. The junior shows the slides of a tiny newborn baby lying in a Moses basket dressed in a white lacy dress and booties- the baby is very thin, and appears to have a small head and prominent ears. She gives a brief history '*this is Megan, this was taken on the day she was born, the first child of unrelated parents and [she] was taken into foster care straight away. The foster mother thought there was something wrong, she didn't smile until 3 months and at 14 months was only just sitting*

up'. The consultant prompts them '*what's the most dramatic thing?*' A junior points to the earlobes; the consultant agrees and describes their qualities '*large, very fleshy and forward facing*'. The juniors suggest a couple of other features such as long fingers and head circumference, but these are dismissed and the other consultant adds that she is '*really quite dysmorphic*'. The juniors fail to add anything and the first consultant breaks the spell, this feature is associated with the SIC 1 mutation, '*they've all got these ears*'. [Meeting 3, case 3]

This is part of the process of teaching the juniors to 'see'. The consultant asks them to describe the child's significant features and then modifies their statements within the language of the specialism. In this extract above, when the juniors attempt to 'see', the consultant re-words their comment so that it fits within the language of the clinic. She also spells out the aspects of that feature that are important for diagnosis, they are '*large, very fleshy and forward facing*'. She thus passes on the skill of seeing and classifying.

The process of knowledge-transmission from consultant to trainee includes ensuring that the junior colleagues '*see a syndrome*': to see one in the clinic is also to be able to classify one. Over 3,000 conditions and syndromes have been described (London Dysmorphology Database (LDDDB); Pictures of Standard Syndromes and Undiagnosed Malformations (POSSUM); Jones 1997), and although some are relatively common such as Fragile X, Cri du chat and Angelman, many are exceedingly rare and a clinician will be fortunate to have 'seen' one in their career. To obtain the skill of assessing and classifying cases, the craft skill of seeing is emphasised.

After lunch the consultant, the Genetic Nurse Specialist and the trainee discuss the cases to be seen that afternoon. The consultant looks in the

medical records of the next case, a ten-year-old boy referred for short stature and states 'it's a Russell-Silver', the trainee responds 'we've been looking for a Russell-Silver' [Russell-Silver syndrome is a pattern of malformations identified simultaneously by Silver in 1953 and by Russell in 1954. The main features are small stature, asymmetry of limbs and a short and/or curved fifth finger and small triangular faces]. We huddle round the consultant as she examines a large pile of family photographs from the medical records and we look at them on the desk. The photographs of a little boy include his birthday party, showing him blowing out the candles on the cake, surrounded by hats, at different developmental stages, newborn, a baby, a toddler and later. The consultant and the trainee discuss his features and decide that the trainee should sit in on this case, she has not seen a Russell-Silver yet and this would add to her expertise. [Clinic 5, patient 4]

This is an important (but difficult) syndrome for dysmorphologists to 'see', but is a common referral for children who have short stature. The diagnosis of this syndrome is not straightforward, there is a marked diversity of features and there are a number of other chromosomal conditions that can resemble Russell-Silver syndrome. Thus, the team scrutinise the images for the 'look' of Russell-Silver. The junior can inspect the visual records of a potential diagnosis at different stages of development and has the important opportunity of increasing her expertise by being able to examine and 'see' a case in the clinic.

Interestingly, the photograph can also be dismissed in favour of the 'eye' of the expert. Despite the importance of the photographs and slides, the examination of the patient in the clinic is often emphasised. Photographs can be out of date and their reliability can be questioned. The lens of the camera can lie; it can enhance, eliminate or distort a 'look' or a feature.

In the team meeting after the clinic, they discuss the case of Sophie, a five-year-old girl who is blind and has severe learning disabilities. Her mother is very young and anxious that she has caused her daughter's problems. The consultant has looked up the child's problems and malformations on a dysmorphology database and although she suggests a number of potential syndromes, they have difficulty finding a classification that would '*fit*'. The MRI scan failed to reveal anything, but the team agrees that the photographs indicate that she looks '*a bit dysmorphic*'. However, the genetic nurse specialist has seen the girl during her home visit and although she agrees that some dysmorphic features are present, she disputes that the child has the 'look' of any of the potential syndromes the team think they can see in the photographs. The consultant agrees that you '*really have to see the children*'.

[Clinic 9, patient 4]

Here a locally well-respected and experienced genetic nurse specialist has the status to pronounce on whether a 'look' that fits a particular syndrome is present. There is an important distinction between being able to identify one or a number of potentially dysmorphic features and the expertise of being able to recognise 'the look' of a syndrome. Implicit within this exchange is trust; the trustworthiness of a diagnosis is often dependent upon who is making the observation.

Oracular pronouncement

The spectacular display of the body or its image is paralleled by the oracular pronouncement of the senior clinician. In this section, we show how clinical authority is displayed through the narration of professional 'experience' and the ability to see and de-code the signs of diseases.

Medicine has been transmitted from teacher to student through encounters that endure, in form, from generation to generation themselves. The clinical 'round' has provided an encounter in which senior practitioners can demonstrate and expatiate on hospital patients since the earliest years of clinical instruction. The round has provided the opportunity for the junior hospital doctor to present a series of patients to the consulting physician or surgeon since the seventeenth century (this is one respect in which Foucault's periodisation of the modern clinic is not universal). The teaching hospital provides a variety of more or less formal occasions in which patients are 'presented' and discussed (Atkinson 1995). The circuits of discourse display and represent 'cases' throughout the modern teaching hospital. Formal grand rounds are paralleled by teaching rounds in which the explicit function of instructing medical students is foremost.

Clinical consultations provide the opportunity for the rehearsal of clinical authority. Consultant physicians do not merely display the classic signs and symptoms of diseases and syndromes; they also display their professional authority and status through a number of rhetorical devices. This rhetoric of clinical authority includes the narration of professional 'experience' and in this context the senior clinician has implicit - but powerful - rights to recount past cases and to ground medical knowledge within a biographical warrant. This biographical knowledge is grounded in the warrant of personal witnessing; an experienced clinician can lay claim to a store of firsthand observations. To have seen a case is to claim direct access to the signs and symptoms of cases and conditions. The phenomenology of the clinic is established by the overriding legitimacy of firsthand testimony.

The consultant discusses the diagnosis of Polymicrogyria with Annabel, an affected teenager and her mother [Polymicrogyria is caused by abnormalities of grey matter of the brain, can lead to developmental delay, speech

difficulties, motor dysfunction of the mouth with drooling, seizures and increased muscle tone]. After describing the structural changes in her brain and discussing the MRI scans, the consultant adds that '*it's not uncommon. It's rare, but I see it quite a lot. Annabel's pattern is typical*'. [Clinic 11, patient 2]

In making a diagnosis, the consultant often commented on the rarity of the diagnosis and displayed her expertise by adding that she has seen many such cases '*I see quite a few and it's not unusual*'. This young woman's subtle features and mild problems fit the diagnosis, but within the context of this specialism she is not unusual and such a seemingly 'rare' diagnosis is common for the consultant to make.

Equally, the claim that one has 'never' seen a syndrome, an associated feature, or a particular kind of clinical presentation, is powerful negative evidence against following a particular diagnosis or a line of argument.

For example, the case of a sixteen-year-old young woman who has been referred with suspected Noonan syndrome. Although the consultant agrees that a number of her problems '*heart problems, learning difficulties and short stature*' do '*fit*' within this classification, she is '*not aware that the facial features of Noonan's include prominent eyes and jaw*'. She has not seen these features and so is unwilling to give a definitive classification until other investigations have been carried out, '*I've not come across the eyes before*'. Although there is no '*definite test*' for this syndrome she decides to take blood and do a platelet count, because low platelets can also be a feature of Noonan syndrome, and the patient is due to have surgery in the near future. [Clinic 2, patient 2]

This patient has a number of the classic features of Noonan syndrome (short stature, learning difficulties, heart abnormalities and a webbed neck). However, her eyes do

not 'fit' the classic case description for this syndrome. Although hypertelorism (widely set eyes), ptosis (drooping eyelids) and downslanting palpebral fissures (the space between the upper and lower eyelids) are all common facial features of Noonan syndrome, this young woman has protruding eyes and so the consultant seeks further evidence with an additional test (platelet count), which may corroborate the diagnosis. A number of features may indicate a syndrome. However, if the consultant sees one feature as in this case, which does not fit in with their clinical experience or is outside the 'classic' description, the classification will be put on hold or other evidence sought.

Yet, even if a clinical feature does not fit within a classification, the consultant can still bring in personal biographical knowledge. Their particular knowledge is given a higher status that can fix a classification.

A woman in her mid thirties has been referred for possible Velo-Cardio-Facial Syndrome [Velo-Cardio-Facial syndrome or Shprintzen syndrome was first reported by Shprintzen in 1978, since when over 100 cases have been reported. The main features are mild learning disabilities, short stature, cleft palate, hearing loss and minor ear anomalies, a prominent nose with a narrow nasal tip and cardiac defects]. The consultant reads the letter of referral, which states the patient has had surgery for a hole in the heart, a cleft palate and vocal cord problems, it also mentions she has '*a very nasal voice*'. After the consultation, she describe the main features the woman has: '*her ears are a little bit small, a pinched nose, typical of the condition...She's also short, her head circumference was normal...she also had marked scoliosis*'. Adding, '*there's little doubt she's got the full house, its surprising given her problems that she's so little sorted out. The scoliosis and epilepsy are difficult, but if*

there's one thing I've learnt, you can get anything with this...She has the full house, so if she doesn't have the deletion I'll eat my hat!'. [Clinic 3, patient 2]

The biographical warrant is evident when the consultant adds that in her experience *'if there's one thing I've learnt, you can get anything with this'*. Thus, the case remains 'classic' despite these additional features (the scoliosis and epilepsy).

Because she has all the features that fit a 'classic' description of the syndrome, the diagnosis is fixed; she has *'the full house'*. This is a classic example of traditional bedside diagnostics where the clinicians can read the pathogenomic signs in the patient's features, she is certain this patient will also have the chromosomal deletion associated with the syndrome (22q), she can 'see' it in her. This occasion is also a display of the consultant's specialist knowledge that other colleagues do not possess. This patient has reached her mid-30's without receiving a diagnosis. She has a huge file of medical records, has had a number of surgical procedures and has been referred to a number of specialists. Only the dysmorphologist can 'see' and classify this classic case.

The dysmorphology meeting is an important site for the enactment of authority. Authority is demonstrated through an ability to 'see' a syndrome and status is enhanced with colleagues by the minimal use of technologies to make a classification. There is added prestige in identifying a syndrome that directs further clinical and genetic investigations, including which test to use and which specialists referrals to make. Thus, one important aspect of authority of diagnosis is acquiring the immediacy of 'seeing' a look, either by reading the visual clues available when the patient is present or by examining photographs or slides.

At a dysmorphology meeting, they review the case of George, a recently diagnosed little boy. The consultant re-caps that *'he was big on all the centiles, he has the pointy chin, deep set nails, we've told mum it's Weaver's'*.

However, her colleague notes that he is '*not typical*' in terms of the features associated with Weaver's syndrome [Weaver and others first reported this syndrome in 1974. The main features are babies who are unusually large at birth, show accelerated growth and skeletal maturation during infancy, mild developmental delay, macrocephaly (large head) and distinctive facial features]. They return to the child's baby photos where the features of the syndrome appear to be more distinct '*his baby photos were really good*'. The consultant tells the juniors, '*I think Weaver's are hard to see as they get older*'. The second consultant adds '*faces change, they suddenly become something or they appear to grow out of something*'. [Meeting 3, case 4]

The most significant features of Weaver's syndrome, accelerated growth and distinct facial features are more visible in babies and infants, but as they get older, the clinicians will have to rely on more subtle features, the 'look', in order to make a diagnosis. Faces can change over time and the skill of the clinician is to see a syndrome through and despite such changes, as the consultant instructs the juniors, '*faces change, they suddenly become something or they appear to grow out of something*'.

This is also associated with keeping people within the clinic; the patient may not have the 'look' of a specific syndrome but there is always the expectation that this may change. The subtlety of diagnosis means that the expertise of clinical geneticists is indispensable- only they can see the signs that indicate the child's problems have a genetic base.

At the dysmorphology meeting, they discuss the case of Joseph. The slide is shown and they exclaim variously that he is a '*gorgeous*', '*very attractive*' '*cute*' little boy. As the consultant notes '*if he was running round Sainsbury's you wouldn't think anything*'. However, she is not happy to discharge him

from the clinic, she points out he has a syndactyly of the toes [the digits are joined], hands that are very soft and hyperextensible [joint laxity] and ‘*sausage-y fingers*’. After they have looked at the slides for some time, the consultant adds, ‘*there’s just something about him*’. [Meeting 6, case 6]

Thus, even though the child has a seemingly ‘normal’ appearance and passes ‘the Sainsbury test’, only their expertise can reveal the subtle signs of an underlying genetic problem. The team are unable to add anything else to aid diagnosis, but they agree to keep him within the clinic, as this may change.

‘Seeing’ also encompasses other senses, not just sight to make a diagnosis. Touch, hearing and smell are also important for decoding the subtle signs of an underlying genetic problem.

During the dysmorphology meeting, one of the juniors gives a history of a recently referred baby. ‘*Small, shows quite autistic behaviour, obsessive behaviour, spontaneous outbursts of laughter, normal chromosomes, normal MECP2....she also has a funny smell, her foster-mother pointed it out, she apparently has it even after a bath...not the musty smell with PKU or the fishy smell, I can’t describe it, but it’s not the clean smell babies have*’. The consultant adds that ‘*the earlobes are significant*’. [Meeting 3, case 3]

Such pronouncements by senior clinicians reflect not just the privilege of experience but also the privilege of the clinical gaze. Here Foucault can guide us. The clinical mentality (cf. Freidson 1970) rests on the charter of the clinician's visual capacity. The physician, the clinical pathologist, the haematologist - these and others can all claim a special capacity to 'see'. The pathologist can see the forms of cells and lesions; the haematologist can read the evidence of a peripheral blood smear or a

bone-marrow aspirate; the clinician can see and de-code the signs of diseases and syndromes. Oracular pronouncements invoke the almost sacred gaze of the clinic.

Genetic technologies

Although diagnosis and clinical classification are to some extent being reshaped by genetic technologies, in this section we show that clinical judgement is still central. Molecular genetic tests do not necessarily enter the clinical process until after the team have reviewed other materials and sometimes they do not enter at all. So although for some cases a result using a molecular test may be viewed as the ultimate proof that a condition is genetic and that its origins reside within an individual's genes, for many of the conditions encountered in the dysmorphology clinic no test is available.

They discuss the case of Sam, a little boy with severe developmental delay. The clinical team are unable to categorise his problems within a specific syndrome, even though he does have some dysmorphic features, he has a '*big forehead*', '*interesting fingers*', '*hairy back*' and his facial features are '*a bit coarse*'. They decide not to carry out any molecular tests but to keep him in the clinic and review him in a year's time '*from a genetics point of view there are no tests we can do, looking at him he doesn't have any of the conditions associated with the specific technologies we have*'. [Clinic 4, patient 3]

Although this child has a number of features that indicate that his problems have a genetic base, the clinician can 'see' that they do not have a genetic cause that can be identified using current technologies. The boy does not have 'the look' that indicates he has the type of mutation or deletion that can be identified by the molecular technologies currently available. However, they decide to keep him among the clinic's patients and to continue to monitor him; technologies may improve in the

future. Thus, not being able to identify a molecular change (or indeed a specific syndrome) does not negate the possibility of identifying a genetic problem in general, even if a specific diagnosis is not available.

Diagnosis and clinical classification are being reshaped by genetic technologies. The use of molecular technologies to identify syndromes has meant that the visual is no longer the only arbiter of classifications. Genetic tests available for some syndromes now mean that the visual basis of some clinical classifications has been called into question.

They discuss the case of a ten-year-old boy who was referred with query Russell-Silver. The consultant does not *'think there's anything significant in his overall appearance to suggest a syndrome'*. However, she takes a range of family photographs for her files. She also takes slides of the front and side of his head, hands palm down, and his feet. After the consultation, she adds that she cannot completely rule out the diagnosis because *'there's been a group of children thought to have Russell-Silver syndrome but you wouldn't recognise them'*. They are only identifiable at the molecular level *'you can get both or part of chromosome 7 from mum'* [instead of one from your father and one from your mother]. [Clinic 5, patient 4]

Apart from the boy's short stature, there appear to be no other visual signs that the consultant can associate with a diagnosis of Russell-Silver. However, she does not dismiss the search for this diagnosis based on her clinical observations, there are a number of chromosomal rearrangements (for example, the maternal uniparental disomy 7 or UPD7 where both or part of both chromosome 7s are from the same parent) that have been associated with Russell-Silver syndrome. Research laboratories are increasingly discovering molecular changes in patient groups, however, the relationship between phenotype (the manifestations of the patient's condition) and the

genotype (the underlying DNA or chromosomal anomaly) is not entirely predictable. As in this case, a chromosomal change is associated with a syndrome, however, there are a group of patients with the same chromosomal change but who do not 'fit' the 'classic' clinical description of the syndrome. Thus, the process of the diagnosis and classification of syndromes is becoming increasingly harder to fix, there are always new molecular changes being identified and linked with syndromes by the research scientists that must be interpreted and adjudicated upon by the clinic.

There is a belief within the team that the technology will improve and the 'fit' between molecular findings (genotype) and clinical features (phenotype) will increase. Blood samples are routinely collected and stored in the expectation that these technologies will improve and provide families with more accurate or appropriate molecular tests in the near future.

At the end of the clinic, the consultant tells the parents of a little boy with Polymicrogyria *'We didn't have a DNA sample from him which we could store. There are three areas being explored so it's only time before we have the technology, it's possible and it could fit with one of those. So that could tell us what caused the problem, but it wouldn't change management'*. They plan to take a blood sample. [Clinic 5, patient 5]

As genetic technologies are introduced for syndrome identification at the molecular level, so clinical classifications may be questioned and refined.

The consultant discusses the case of a little boy with severe developmental delay with his parents. She discusses the diagnosis of polymicrogyria. [Polymicrogyria is associated with developmental delay, seizures and decreased muscle tone which delays development of infant motor milestones such as head support and sitting. Later this is evident from a slumped sitting posture, late walking and an abnormal gait]. The consultant adds *'one of the*

questions we were going to ask again in terms of Polymicrogyria we're learning a lot, we can distinguish the types, we think he has the common one, epilepsy, drooling and gait abnormalities. Things are moving and we could look at other things, we could look at chromosome changes, 21 and 22, we could look at changes there but we didn't know enough about it to say, but [its] worthwhile excluding it.' [Clinic 5, patient 5]

The syndrome Polymicrogyria is being re-defined by molecular tests and re-negotiated by the clinic. Although there is a 'common' type, the main features of which are '*epilepsy, drooling, gait abnormalities*', there are also other sub-categories associated with changes on chromosome 21 and 22 that have been identified. Thus, for some syndromes, clinical diagnoses are developing a more subtle taxonomy in light of genetic laboratory work.

The remarkable rate of growth in new genetic techniques and the identification of a genetic basis for a wide range of conditions have had considerable implications for clinical medicine. It is now possible to use specific genetic tests to identify a number of conditions previously classified only in clinical terms. It would be wrong, however, to assume that there is a linear evolutionary sequence at work here - from clinical perception to laboratory testing. In the four decades since the first genetic investigations, genetic science has progressed rapidly, so that much smaller molecular and chromosomal changes can be determined. There are now potentially so many genetic alterations that can be detected that the element of clinical judgement is not so much being lost as re-directed into deciding which of the possible laboratory tests should be applied in the assessment of each case.

These processes are not stable or immutable. Even when new molecular technologies are used, clinical judgement is still central; their use is dependent upon

the expertise of the clinicians. Tests are not used indiscriminately; indeed a demonstration of the expertise of the clinician is in using such tests appropriately.

After examining Anthony, a boy with learning disabilities and behavioural problems, the consultant explains to his mother and grandmother: *'I'm interested in movements, he was really good up there [examination table]. I'm interested in movements because a new gene has been identified, these boys have learning difficulties and some funny movements...we're setting up this test [here] and we could add Anthony if you would like to go down that route...that's the only idea I have at the moment...I may have others in the future'*. [Clinic 1, patient 1]

Here the consultant considers a new genetic test to identify a molecular change, which is associated with learning difficulties and *'movements'*. Only a skilled clinician can 'see' these subtle signs and associate them with the relevant genetic test. The tests are dependent upon the skill of the clinician to align the 'sign' in the patient accurately with the appropriate molecular technology.

Even if a syndrome can be identified at a molecular level, the subtleties of the clinical classification are central to diagnosis. A negative or a positive test result can equally be dismissed or enrolled into a classification, dependent upon the other evidence available and whether it fits with what the clinicians can 'see' in the patient. The subtleties of classification and 'seeing a syndrome' are still of key importance.

The team discuss the case of William, a teenager with autistic spectrum disorder. The consultant considers the risk to his brother of having a similarly affected child and the likely pattern of inheritance. Adding that they would be unlikely to find any evidence using a genetic test, it's *'not unusual to have this type of pattern in other family members so I think genetic tests in William would be normal'*. They decide not to take blood. [Clinic 5, patient 2]

The team often pronounced that they ‘knew’ a molecular change would or would not be present in an individual before they had received the test results, they could see its external expression in the individual’s features. In the case above, they decide not to carry out a genetic test because they do not believe it would provide additional information and the clinical diagnosis and associated familial risk for the brother are fixed.

Expertise is similarly displayed in identifying the signs and symptoms that would find their expression in a specific molecular test.

The team discuss the case of a ten-year-old boy referred with possible Russell-Silver syndrome. The consultant dismisses this classification for a number of reasons and the trainee agrees that he doesn’t have ‘*the face*’. However one feature, his ‘*marked 5th finger clinodactyly*’ [a curved fifth finger] leads her to believe that a new molecular test may provide a result, ‘*I think he’s a good candidate for UPD7’s [both or part of both chromosome 7s from the same parent, in this case the mother] he is short and has marked clinodactyly*’.

They agree to take blood and run the test. [Clinic 5, patient 4]

Thus, an important skill is knowing when a patient would be likely to get a positive test result. In this case, the team recognize the chromosomal change is likely to be there, they can ‘see’ it in his features. Although this boy does not have ‘the face’ that fits the classic features of the syndrome, the clinicians can identify the subtle sign (the curved fifth finger) that may be associated with a specific genetic test.

If the genetic technologies fail to reveal or identify the predicted chromosomal change, the team do not dismiss their diagnosis; rather the rhetoric of improving technology is employed or they transfer their search to other sites of the body such as skin, blood or brain.

In the team meeting they discuss the case of Jacob, a little boy who had '*got lots of problems, he's going to be adopted, lots of social issues*'. The consultant says that she knows there is '*something in the chromosomes*'.

However, '*we haven't found it yet, so we're looking at the skin*', they were unable to locate the chromosomal change in his blood. [Meeting 1, case 5]

The consultant can 'see' the chromosomal change in the child. Thus, the inspection of the body and recognition of their 'look' that fits a classification can lead to a negative genetic test result being dismissed. Rather than dismissing the clinical diagnosis, the team continues to look for the genetic change by moving to different sites of the body to confirm the clinical diagnosis, this time in the boy's skin. The embodiment of medical knowledge and authority here includes the competence to 'see' in a particularly adept and privileged way. The observation and recognition of classic signs and characteristic appearances are among the ways in which medical authority and disease entities are simultaneously constituted (Canguilhem 1989).

Discussion

We have described some deliberations of the clinic to illuminate how *in practice* genetic science informs clinical judgement, contributing to the configuration and re-configuration of syndromes and cases. We have suggested that contemporary dysmorphology can be understood in terms of long-standing forms of medical knowledge, medical representations and medical discourse (King 1982).

Notwithstanding the new forms of technology provided by genetic science (Casper and Koenig 1996; Keating and Cambrosio 2001), 'the clinic' still asserts its symbolic and functional power: the 'gaze' of the clinician and the clinician's warrant of personal knowledge still exert their influence.

The work of the genetics services includes the ascription of specific named conditions to patients. This involves the assembly of a clinical description, including the characterization of the patient's physical appearance, including - but not exclusively - the appearance of the head and face. Clinicians decide whether appearances are 'normal' or 'abnormal', and - if abnormal - whether they correspond to a dysmorphic clinical entity. The classification itself has a degree of flexibility in it; we are here studying the process of *making and re-making* syndromes as well as their description. As Canguilhem (1992) classically pointed out, and as Keating and Cambrosio (2003) have more recently affirmed, the pathological is no mere extension of the biological, but is constituted by distinctive, shifting configurations of technique, judgment and representation. The clinic is a site in which entities are assembled, and is obdurately resistant to statistical or biological reduction. We are not witnessing a simple reductionist 'geneticisation' of medical knowledge in this context (cf. Hedgecoe 1998, 1999, 2003; Kerr 2000, 2004). There is no single hierarchy of knowledge-types. There are, however, hierarchical relations of expertise. Locally, nationally and internationally specialists in dysmorphology are recognised by their professional peers as having personal and sapiential authority in recognising syndromes, and adjudicating cases. We have therefore documented what we have called 'the spectacle of the clinic', emphasizing the performative and visual aspects of clinical work and inference in the adjudication of dysmorphia. We propose that the contemporary practice of dysmorphology displays the intersection of two organizing principles in the constructing and mobilization of medical knowledge. On the one hand, it is a site in which new medical technologies are employed and interpreted. On the other hand, it displays the long-standing features of clinical medicine. We are here witnessing, therefore, the intersection of the 'old' clinic and the 'new'.

This analysis of the performance of dysmorphology has stressed the visual culture of the clinic (cf. Cartwright 1995; Atkinson 1995; Kevles 1997). The inspection of patients' appearances and the uses of visual representations in assembling a clinical dysmorphic description highlights the spectacular display of clinical work. Dysmorphology thus furnishes a graphic exemplar of the visualisation of medical evidence in the construction of clinical entities (cf. Kemp and Wallace 2000; Heath 1998). The visualisation of 'family' through the family tree and family photographs are among the devices that render legible the families and their shared characteristics (cf. Nukaga and Cambrosio 1991; Gibbon 2002). Likewise, the scrutiny of individual patients through slides and other representations creates the occasion for clinicians to display their acumen, experience and trained eye in assembling descriptions of typical abnormalities, and to adjudicate when perceived characteristics are adequately 'syndromic'. As a specialty, dysmorphology provides a rich and developing *nosography*. As we have indicated, the categorization of syndromes is not static. The classification and description of dysmorphic syndromes are subject to modification. This is, therefore, a nosography-in-the-making for some conditions at least. Dysmorphology has thus furnished us with a prime opportunity to document the processes of medical classification as it occurs (cf. Bowker and Star 1999). We suggest that a broad historical and cultural pattern can be traced that brings together the *spectacular display* and the *oracular pronouncement* as long-standing (although by no means immutable) features of medical knowledge and the importance of a deeply entrenched visual and oral culture in the creation and transmission of medical knowledge.

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