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ABNORMAL APPEARANCES:
INSPECTION, DISPLAY AND THE CLINIC

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SUMMARY

ABNORMAL APPEARANCES: INSPECTION, DISPLAY AND THE CLINIC

We provide an examination of the field of dysmorphology, a clinical speciality that in its current form combines a long history of inspection and display with the identification and representation of associated underlying molecular changes. The recognition and description of abnormal appearances is thus increasingly accompanied by genetic and other molecular investigations. Our analysis draws on our long-term ethnographic engagement with a UK clinical genetics service and the work of two clinical genetics teams within a regional teaching hospital. We document the intersection of genetic science with clinical work to suggest that while molecular testing often identifies the genetic basis for unusual appearances and abnormal development, it does not fully supplant clinical apperception and interpretation. The two modes of knowledge – the clinical and the biomedical – co-exist in the work and the discourse of dysmorphology practice. The contemporary dysmorphology clinic thus encapsulates the epistemological systems of modern medicine, grounded in the clinical gaze and on the classificatory systems of classic nosology. Within such a system of clinical knowledge, the ‘monstrous’ does not escape the boundaries of knowledge. Monstrous appearances are accommodated and domesticated within the classificatory systems of normal medicine.

Key words: Diagnosis – Classification – Genetic Syndrome – Genetic Testing – Children – Ethnography

Introduction

Medical monstrosities have a long history¹. They also have a long history of display, as specimens or objects of curiosity². Contemporary medical discourse may no longer have a place for such creatures. But the clinic remains a site for the spectacular display of abnormal appearances, and for a florid system of classifications, whereby the particulars of unusual appearance and abnormal development are inspected and described³. The clinical specialty of dysmorphology provides opportunities to analyse such processes in action, in the contemporary clinic. We therefore examine the spectacular presentation, collection and representation of patients, their bodies and their identities within the specialist clinic. We trace the visual culture in the dysmorphology clinic. The specialist clinic is a site in which we can observe the complex, practical work of diagnosis and of fitting a patient, in this case, a child with a suspected genetic syndrome. We consider this diagnostic work in terms of the spectacular display of the clinic, the collection of cases and examine the expert's competence in 'seeing' cases and interpreting visual representations. Notwithstanding the multiple developments of biomedical science, including new genetic technologies, dysmorphology retains classic elements of the clinical gaze. It furnishes a rich nosography of clinical descriptions, and a classificatory nosology of abnormal development and appearance.

Dysmorphology and Abnormal Appearances

Contemporary dysmorphology stands at the crossroads of the old clinic and the new technologies of genomic science. It preserves many features of earlier forms of physiognomy and iconography; however, in recent years, it has also been subject to increasing technical change and 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

technologies⁴. Thus clinical dysmorphology parallels other clinical areas and represents an important case at the intersection of clinical medicine and genetic science.

The term 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, sometimes including delayed intellectual maturation⁵. These patterns are associated with abnormalities but are not necessarily abnormal in themselves. However, particular patterns of physical features have come to be associated with underlying systemic abnormalities such as heart defects, or delayed intellectual development. Patients are mainly babies, children and teenagers or young adults. For example, Rett syndrome became more widely known in the early 80's following the publication of first English language account (Rett, 1977)⁶ and there have been a number of attempts to develop consensus on the key criteria required for a diagnosis of Rett syndrome⁷. Classically, affected girls (this syndrome is extremely rare in boys) are described as developing normally until they reach 6 to 18 months old when their development appears to slow down. This leads to the start of a period of regression, which may cause a sudden and dramatic loss of skills, although this may have a subtle onset. Regression is often accompanied by social withdrawal and leads to a loss of skills, particularly speech with any prior words they have learnt usually permanently lost and typically they become unable to use their hands purposefully and girls affected by classic Rett syndrome are profoundly intellectually disabled. Progressive physical problems can also develop in later stages of the syndrome, including scoliosis and behavioral problems. Rett syndrome is not associated with a strong dysmorphic facial 'look'; however, the key physical or behavioral feature is the striking repetitive hand movements and atypical breathing patterns that typically develop following the period of regression.

Dysmorphology also retains a classic, modern medical preoccupation with classificatory systems and the proliferation of named clinical entities. Typically, the features of abnormal appearance and their fine-grained description provide the basis for identifying a variety of named conditions. When patterns are deemed to have reached a level of regularity across different cases they are defined and named as a syndrome. There are several thousand named dysmorphic syndromes currently held within international clinical databases and textbooks. The majority of syndromes have been identified as having a genetic basis, which are either single gene defects or chromosomal disorders. Chromosomal abnormalities are spontaneous, *de novo* occurrences. When this is believed to be the cause of a child's condition, the risk of recurrence within the family is assessed as being low, particularly where no abnormality is present in a parent. However, some syndromes are familial conditions as a consequence of an inherited genetic defect. If this is the case the clinic will attempt to identify the underlying genetic constitution, and provide families with an estimate of the likely risk of recurrence in future pregnancies. The recognition and description of abnormal appearances is increasingly accompanied by genetic and other molecular investigations.

The specific features that distinguish clinical dysmorphology include: the recognition and classification of specific patterns of facial and other physical features; ongoing classification based on clinical diagnosis and examination; use and interpretation of molecular tests in diagnosis and clinical classification; decision-making and assessment distributed and between different experts (including scientists and clinicians) at local, regional and national level; the occasional introduction of new clinical categories and diagnostic labels. Thus, an examination of the field of dysmorphology provides a speciality that displays the interaction of genetic technologies and clinical judgement. In its current form it combines a long history of inspection and display with the identification and representation of associated underlying molecular changes.

We ourselves have already documented some of the devices through which the dysmorphic appearance is constructed, and how genetic syndromes are constituted⁸. Others have examined the paediatric and genetics clinic to examine its impact on inheritance and responsibility⁹, identity¹⁰, and the role of visual technologies to display and interpret molecular findings¹¹ in the context of the diagnosis and classification of dysmorphic genetic syndromes.

More widely, the experience of parents who have a child with a disability or spoiled appearance has been a focus for research since the early 1970s¹². We have examined parental perceptions of stigma and the sentimental and moral work performed in the genetics clinic, which are issues of particular significance in the context of dysmorphology¹³ and there exists an extensive literature examining parental perceptions of “courtesy stigma”¹⁴ and the stigmatized identities of children with a disability. With a focus on conditions such as craniofacial disorders¹⁵, Down syndrome¹⁶ and obesity in children¹⁷. Additionally, there are a number of studies examining families with “discreditable” members, where behavioral characteristics, although not immediately apparent, are potential threats to children’s – and parents’ – identities. These include disorders of developmental coordination¹⁸ and epilepsy¹⁹. Studies have also examined the coping mechanisms of parents with a potential identity spoiled by association²⁰.

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 wrote vividly on the clinical ‘gaze’ (*le regard*) in the development of the modern clinic²¹. He suggested that

in 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.

Foucault is right up to a point, but he is wrong – and interpreters are wrong – If they focus too narrowly on his account of the clinic. Undeniably, modern medicine has rested on a claim to privileged access to clinical perception. The modern clinic has indeed treated the patient's body as a site of privileged perception, and the bedside as an equally privileged space for the exercise of a distinctive clinical perception. However, Foucault's insistence on the specificity of his analysis - the particular institutional and epistemic conditions of Paris at a particular historical juncture – must be taken into account. Moreover, his preoccupation with disjunctures and transformations leads him to underplay the long historical continuities in medical perception and the forms of medical display.

Sociologists of biomedical knowledge have attempted various periodisations, to account for major changes in technologies and the organisation of work. An influential example is provided by Clarke, et al (2003)²², who develop a contrast between "medicalization" and "biomedicalization". The former corresponds to medicine based on local clinical practice, grounded in "cases", and dependent on individual practitioners, while the latter corresponds to evidence-based medicine, based on scientific and technological innovations, such as molecularization and genomics, and distributed expertise. Our argument is that such periodization, while not wrong, is potentially misleading. "Modern" medicine has not been supplanted by biomedical-

ized modes of perception and practice. Rather, we should think of a palimpsest of knowledge-types and practices. Biomedicalization and its techniques do not replace more classical classifications of diseases, syndromes and conditions. Contemporary technologies do not supplant the clinic as a site of practice, nor do they overturn the distinctively clinical work of observation and inference. We illustrate this contention with reference to the clinical specialty of dysmorphology, the investigation and classification of abnormal development and its manifestations in named syndromes. Our study is, therefore, an aspect of the “history of the present”, documenting the intersection of genetic biomedical science and clinical work. While we do not subscribe to Foucault’s historical account of the emergence of modern medical knowledge, the notion of the clinical gaze is a valuable sensitizing concept, in drawing attention to the distinctive work of the clinic.

Presentation and Representations

The social *forms* of the spectacle are various, and have their own *longue durée*. The early modern anatomy lesson²³ is a classic case in point. The classic 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”²⁴. The anatomy lesson was translated into artistic representation through the *écorché* (flayed) figure that featured in the anatomy lessons of the art academies. The body became transformed into a spectacle through its representation, and the circulation of such representations for didactic, diagnostic and aesthetic purposes.

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. From fine-art anatomical drawings, to the engraved

plates of anatomical atlases, to wax anatomical figures, through to modern imaging technologies²⁵. The convergence of art, literature, social science and medical science on the representation of the body has in recent years given a new currency to a much older set of traditions and artefacts. The spectacle of the body and the spectacle of the clinic have a long and complex history. Sometimes the patient has been enrolled as a 'stooge' in the clinical display and attended the theatre for a clinical lecture or display. Sometimes the patient has been an accomplice, or even a star turn, for example, some of the most famous accomplices in the history of the spectacular are Charcot's performing hysterics at La Salpêtrière. Sometimes the "patient" is absent, represented only in fragments such as frozen sections or other specimens presented at clinico-pathological conferences and presentations. Thus, the patient is interpreted and translated through the assemblage of shards of evidence. The recognition and adjudication of pathology in these 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.

There is also a long tradition in which patients' appearances are captured. 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 rich mechanical means that complemented and then supplanted fine-art traditions in the representation of physiognomy.

The discipline 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. Appearance has long been thought to reveal the inner character of the person and as Kemp and Wallace 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”²⁶. Artists’ exercises have included the visual equation of human and animal types in capturing character traits, the portrayal of racial types and the depiction of the insane. 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 most famous of the early exercises in the depiction of character was the work of Lombroso, who equated criminal and physiognomic types. The photographic family album and the family tree were transformed into the visual display of physiological and phrenological semiology. In the same way, Francis Galton in the United Kingdom combined the resources of photography with anthropometric techniques to classifying human types (as with Lombroso, his early dataset was Home Office portraits of convicted criminals.). Although Lombroso and Galton were interested in the classification of types, and in the correspondence between appearance and character, they were also interested in the hereditary transmission of character from generation to generation.

The depiction of the insane was also a major site for the photographic recording of appearance and pathology. Asylum patients were photographed in abundance from the middle of the nineteenth century, for example in England notably by Hugh Welch Diamond, one of the founders of the Royal Photographic Society. More famously, an extensive photographic record supplemented Charcot’s displays of patients at the Salpêtrière. In collaboration with photographers Bourneville, Regnard and Londe, Charcot established a photograph-

ic unit at the hospital and the publication of *New Iconography of the Salpêtrière* enshrined a voluminous record of the hysterics, cataleptics and other inmates.

The modern clinic is now suffused with images of patients as well as 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. It can be found in the presentation and discussion of the dysmorphic patient and the adjudication of dysmorphological nosography. Such presentations are also sites for the enactment of oracular authority by genetic scientists and clinicians.

Teaching rounds, grand rounds and the other circuits are ritualised occasions for the affirmation of professional seniority. There is a discursive hierarchy in the rituals of the round, junior staff and students present patients by recounting their history, summarising their current hospital admission and enumerating clinical findings, while their senior colleagues pronounce and adjudicate. However, the “round” is no longer restricted to a physical tour of inspection of the hospital ward and its inmates. Rounds do still take the form of sacred progresses from bedside to bedside²⁷, but many events that are designated as rounds - or which have equivalent functions but without the label - take place round a table. Patients are presented and clinical findings discussed in a setting physically removed from the ward itself and in consulting specialties, the individual patients may be elsewhere in any case. Cases are discussed *in absentia* and are based on visual and laboratory evidence alone. The dysmorphology clinics and professional meetings that are the focus of this paper are of this type.

Dysmorphologists are, therefore, preoccupied with the recognition, adjudication and classification of *abnormal appearances*, usually in young children, who display physical characteristics of abnormality, often accompanied by cognitive impairment. Their work involves the attribution of an extraordinarily wide and diverse range of *types*, usually in the form of named syndromes. While molecular testing often identifies the genetic basis for unusual appearances and abnormal development, it does not fully supplant the exercise of clinical apperception and interpretation. The two modes of knowledge – the clinical and the biomedical – co-exist in the work and the discourse of dysmorphology practice.

While medical scientists and embryologists stress the value of mutants for our collective understanding of normal development²⁸, cultural analysts have frequently invoked the monstrous and the abject in ways that are essentially metaphorical²⁹. In a typical move, the essays by Law (1991) and Star (1991) treat the monstrous in terms of contested technologies³⁰. But they have little to say about technologies of investigation and classification of ‘monstrous’ bodies themselves. Indeed, Hughes (2009) – writing specifically about disability studies – suggests that most cultural analyses are about words rather than carnal bodies³¹. In our analysis, on the other hand, we deal directly with the carnal reality of abnormality, and the embodied work that medical specialists engage in as they describe and classify dysmorphic appearances. To that extent ‘the clinical gaze’ is not a metaphor, or an ideal-type. It is a literal description of that form of work.

Methods

As part of our long-term ethnographic engagement with a UK clinical genetics service, we followed the work of two clinical genetics teams within a regional teaching hospital. The first was a clinical genetics team (consultant, trainee and specialist nurse) specialising in the diagnosis of genetic syndromes over a period of nine months, from

2002 to 2003³², the second, a team of genetic and other clinical specialists (paediatrics, neurology, psychiatry) who were taking referrals of suspected cases from across the UK and specialising in the diagnosis and assessment of one genetic syndrome (Rett syndrome) over a two-year period from 2004 to 2006³³. The teaching hospital serviced a wider regional area and across the two projects we observed consultations within clinics based across five local satellite hospitals. These are very different kinds of consultation from the fleeting encounters characteristic of most primary care settings, with the average length of time allocated to each consultation was one hour. In addition, we observed local professional dysmorphology meetings where cases were presented and discussed and a large number of less formal encounters between professionals were observed. Given the nature of these conditions, cases of dysmorphia are overwhelmingly children, whose physical and mental development have been perceived as problematic. Our examples draw upon detailed fieldnotes taken (by Featherstone) during and immediately following each period of observation and include segments of near-verbatim text. This project was approved by a UK Multi-centre Research Ethics Committee. All names have been changed to preserve anonymity.

The Organization and Division of Genetic Work

In examining the work of the clinic we do not refer simply to a focus on the core encounter between the practitioner and the patient or “client”. Nor do we mean we are examining the work of one physical, organizational location within the healthcare system (such as a ward or hospital). Rather, we refer to a wider constellation of settings, clinical staff, and occasions, at which clinicians and others engage in characteristic forms of medical work. Although talking to and examining patients is a key feature, this work also involves examining medical records, and inspecting evidence (such as test results and laboratory values, images and scans), regular professional meetings (such as the

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clinical case conference), training encounters (such as the clinical lecture or the teaching round) and other meetings with the patient or family (specialist nurses within the service often carry out home visits to initially assess patients and families). In specialist services such as this, what is often called a “clinic” involves professional discussions that incorporate several of these encounters and layers of work. These clinics focused on the assessment and classification of children with profound intellectual disabilities thought to be genetic in origin. These cases are usually not familial, but are chromosomal disorders that occur spontaneously (*de novo*) in an individual. The striking feature of these clinical encounters was the close physical inspection of the child’s body to look for signs that they might have an underlying genetic disorder or syndrome. Talking to the clinical team we had been initially led to believe that genetic technologies was taking over the field and rapidly entering and transforming the work of diagnosis and classification. Yet despite these technical developments, many genetic abnormalities that entered the clinic could not be identified through the use of either molecular or cytogenetic (chromosomal) tests. We were particularly intrigued by the clinical team’s use of the more traditional classificatory techniques of inspection and adjudication and the reliance on the expertise of the clinical specialist to “see” a syndrome; we wanted to follow this work more closely. The work of the clinic is conducted, in large measure, through the inspection of the child. Such work invokes the *visual culture* of the clinic. In the contemporary clinic, appearances are correlated with the findings of molecular biology. The organ is replaced by the molecule, but the gaze remains central.

Inspecting the Child

The diagnosis of syndromes relies on the traditional clinical skills of examining the body for signs of underlying pathology. In general within dysmorphology clinics, the detailed inspection of a child’s ap-

pearance persists. It can be found in the presentation and discussion of the dysmorphic patient and the adjudication of dysmorphological nosography. Each child who enters the clinic is subject to a close and detailed observation by the clinical team during every consultation in order to read and interpret significance of each feature in the context of the potential syndrome. The following case summary from our fieldnotes illustrates how the visual culture of the clinic is embedded in the routine work of physical touching. Here the visual inspection is accompanied by a haptic (touch) exploration. Once the child's history has been taken, the team member leading the consultation typically knelt down in front of the child and initiated the examination by closely scrutinizing each feature of their face and their head (including hair and ears). This examination involved a lot of touching of the child's body by the consultant, using their hands to inspect and feel each feature. The child's hands were also closely examined by looking at both sides and each finger in detail, as were the feet with shoes and socks being taken off. The child was often undressed to her underwear, her limbs examined and her reflexes tested. At this stage the child's back and spine were closely examined for any curvature (scoliosis is a feature of some syndromes) and the consultant used their hands to feel along the spine. At this point it was usual for other members of the team to join in and comment on this feature:

In this case, they were examining Tamsin, a 4-year-old girl with a query diagnosis of Rett syndrome attending with her parents, and as the examination drew to a close the consultant announced to the room "she doesn't have typical Rett feet" (small feet although not a central feature, are associated with Rett syndrome). None of the other team members responded to this statement, but it becomes part of the process of establishing whether a feature is within the normal range, or a sign of the underlying syndrome.

The clinical team search in the visual presentation of the child for ways to distinguish physical features, what is normal and what is

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abnormal and also which features are part of the potential syndrome being considered and which should be dismissed or are not 'typical' for this syndrome and may lead to this syndrome being rejected and an alternative diagnosis being considered. They map the child's body to identify and align it with features associated with this syndrome. Clinical appearance includes more than just the inspection and assessment of the physical features of the body. Behavioural features are often associated with specific syndromes: observing the child's conduct in the clinic is also significant. The clinical team spent a lot of time during each consultation focussing on the child and watching them:

In this same consultation all members of the team concentrated on Tamsin, saying "Hello" to her as she circled the room. This child was very active and walked around the room wringing her hands. All members of the team were very much concentrating on her and on one level this appears to be a celebration of this child, however, this also has a clinical function; the team are searching for evidence. Typically they were observing her gait, the way she walked and her co-ordination and also the child's gaze, whether she walked up to individuals, looked at people and focussed on their faces. At a later point in the consultation Tamsin was given a biscuit and managed to drop it and spread crumbs all over the floor. This was not treated as a problem that must be cleaned up and managed, but became a focus for the team who all watched her intently, keen to see how she dealt with this incident.

Again, this sentimental work also has an underlying clinical function. The clinical team were looking for ways to distinguish her behavioral and 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 or behaviour deviates from the normal range and if it fits within the established features of this syndrome.

Such features are not examined in isolation but are also compared with those of the child's immediate family – it may be outside the normal range, but a benign feature within this particular kindred. In this case,

the child's ears and hands are evaluated and commented on and it is important to establish whether such a feature is familial because if not, it may be a clue to the underlying syndrome. Here the clinician is reporting initial findings to her colleagues at a clinical meeting:

I started looking at Jessica initially and thought oh yes she's a little bit coarse and a little bit and a certain something about her that was quite hard to quantify, but you know didn't look, you know, perhaps in the normal range if you like but then when I was examining her every time I found something, I kept looking at her and then looking across at mum and thinking, oh yes that's mum, so I was looking at her eye shape and thinking oh her eyes are quite small and quite deep set and so were mum's and then I looked at her nose and thought oh, Jessica's nose is quite long and sort of the tip's quite pronounced. And then I looked at mum and thought well mum's got a long face, mum's got the long nose as well she was very much her mother's daughter, you could really see that.

She goes on to explain that whilst the child's features are associated with Rett syndrome they may also be associated with another syndrome that is similar to Rett - Angelmans syndrome. She brings together the features she found most interesting (ears and hands) with her teeth into a general category of bone structure and links this to two other girls with a diagnosis of Rett syndrome she had recently assessed. This example illustrates the practical reasoning that ties the close observation of abnormal appearances, the descriptive categories employed, and the classificatory system to which abnormal developments are attributed.

Identifying the abnormal

The children who attended these clinics had dysmorphic features of varying severity, some of which related to the face or head. Some 'abnormal' physical features may be perceived as giving rise to a spoiled appearance: for example, craniostenosis (an enlargement of the skull). Paradoxically, some equally 'abnormal' features can also

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be extremely attractive, for example, children with elfin features, triangular faces and small stature are associated with a number of syndromes.

The team routinely described children attending the clinic in terms of their physical attractiveness irrespective of the apparent severity of their dysmorphic features. For example, a young boy with suspected Russell-Silver syndrome (the main features of which are small stature, asymmetry of limbs, a short and/or curved fifth finger and small triangular faces) is described as “gorgeous” and “a little charmer”; a little girl at risk of inherited cardiomyopathy (a disease of the heart muscle that can lead to sudden death) is a “gorgeous little girl”, and a child with 22Q (associated with a deletion of the long arm of chromosome 22, this syndrome has variable dysmorphic features consisting of a round face, almond-shaped palpebral fissures, bulbous nose, malformed ears, hypotonia, short stature, learning disabilities, and other anomalies) is “very sweet”. The clinical team often explicitly described the child’s features to parents in a positive way, using adjectives such as “pretty”, “handsome” and “gorgeous”. This extended to the examination of some children with severe physical abnormalities. In the example below, this young child has Goldenhar syndrome (hemifacial microsomia), his features are clearly asymmetric, and he has dysplastic ears (low and set back), large auricular tags (skin tags near the ear), epibulbar dermoid (ophthalmology problems) and mild facial weakness on his right side. The consultant concludes her examination by declaring that he is “gorgeous”. She appears to play down the severity of his abnormalities even in the face of parental insistence that his physical malformations are severe:

Consultant Geneticist: His asymmetry is not that marked.

Mother: The position of his ears is quite different.

Consultant Geneticist: [To the child, holding his head in her hands.] You don’t look too bad at all, in fact gorgeous!

However, even when children did not display any clear physical features that would mark them out from others, this does not mean that they are dismissed from the clinic. The clinicians display their expertise by seeing these subtle signs that a normal eye would not see. The consultant below notes that this child would pass the ‘supermarket test’, meaning that they would go unnoticed in normal social life. However, only she can see the subtle signs and even though she is not able to provide a diagnosis at this appointment, he stays within her clinical caseload and she considers further tests to identify the underlying cause of his problems:

Consultant: If he was running round Sainsbury's [UK supermarket] you wouldn't think anything... we've excluded FRAX, Prada Willy, and worth doing a bone age to see if he's overgrown, likely to be looking at something different. There isn't really a phenotype for amphetamines. It's sad but I'm not happy to write him off.

SpR: He's cute.

Not all unusual appearances are necessarily attributed to an underlying syndrome. In the following clinical encounter, the clinician is very excited to find that this young boy not only has hairy arms, but also has a ‘hairy back’, which she examines closely. His mother is also present, and takes part in the discussion, being drawn into a comparison with other members of the family who are not present:

The consultant examines the boy's arms, "He's got hairy arms". Mother replies, "Yes, and a hairy back". Dr, "Ooh, let's have a look at that". She rolls up his vest to have a closer look; you can see a fine down of dark hair running down his spine. Mother: "His father is very hairy". Consultant: "Yes he has a hairy line down his back [...]. Does your husband have thick hair as well?" Mother: "Yes". Consultant: "You do as well?". Mother: "Yes, and my daughter". The consultant takes the boy's shoes and socks off. She looks at his knees, ankles and toes and looks between his toes. She takes his trousers off and gets him to stand up. She examines his thighs and

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legs and feels his stomach. She pulls open his pants and looks quickly. Consultant: "A little peek down below, all OK [...]. Have you got any pictures of your daughter?". The consultant goes to her desk and opens an envelope contained in the medical records and examines the slides within. The child goes to the desk and looks at one of the slides. Consultant: "Can we take an up-to-date picture?". Mother gives the consultant a small picture of her daughter that she carries in her wallet. The consultant compares the photo with one of the slides, she then hands them to the consultant and then to me. The daughter is extremely pretty with big blue eyes, strong cheekbones and very long thick hair. Mother: "They look similar". Consultant: "Yes they do, big blue eyes and thick hair".

The clinician is looking for ways to distinguish physical features and establish 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. Importantly, in the context of the wider family, the father who is "hairy" and the sister who looks similar and has the same "big blue eyes and thick hair", the consultant decides it is likely to be familial rather than a trait associated with an underlying syndrome. However, the consultant files the family photograph of the daughter provided, and takes an up-to-date photograph of the boy for future reference, features change over time and may become significant later as the child matures.

The key features the clinical team are looking for are often subtle and elusive. I observe the clinician assessing a child at home prior to her attendance at the clinic. Jessica, a five-year-old little girl, is tiny and frail, and she has a gastrostomy tube; she has no speech and has daily seizures and long screaming fits. As well as taking a history from her mother, and recording the child via photographs and video, the clinician also examines the child's features. During the journey back to the office she describes what she has seen. Importantly, she does not focus on the highly visible and observable features of this child's problems, but on the child's ears and fingers:

It's quite interesting to see things like her ears. Jessica's ears are sort of... a bit too complicated, a bit too many faults, a few too many creases, the ears are quite long and sort of quite pointy at the top. And then the earlobes are sort of very much attached to the ear but, you know not connected to the side of the face at all; there's virtually no connection whatsoever between the earlobe and the side of the face so it almost looks like the earlobes are hanging on by a thread... The fingers were really quite short. She had tiny little hands except it was all tiny because of the fingers, the palms were fine, it was just because all the fingers were a bit short and then, you know we talk about clinodactyly (curving in of the little finger towards the thumb), that was quite marked in both hands. It was just these fingers were quite little but it wasn't any one part of the finger was shorter than the rest of it, it was that all the fingers were generally a bit short.

The visual culture of dysmorphology is central. The expert must develop competence in 'seeing' cases and interpreting visual representations.

Butterfly collecting: capturing the syndrome

The collection of types and cases is an important part of the process of developing expertise. The most prominent visual technology utilised in the process of adjudication and diagnosis of syndromes within the general dysmorphology clinic is the photograph (Featherstone et al, 2004). There are two types of photograph employed by the clinical team, family photographs and slides taken during the clinical consultation. Family photographs typically featuring special occasions marking celebrations are collected at the initial home visit (this is either done by the Specialist Nurse or Genetic counsellor) and are used to examine the child's features, but also other members of the family for signs of an underlying syndrome. Occasionally family photographs of the child when they were younger were collected and scrutinized by the team, who were often looking for stages in their earlier development when the "look" of the syndrome could be seen more clearly. Where photographs are available they become part of

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the evidence used to make a diagnosis and of initiating the process of identifying the child as having a genetic syndrome. During the clinical consultation, photographs were routinely taken of every child and they typically featured the front of the face, pictures of each profile and close-ups of feet, hands and any other interesting features the clinician thought may be significant such as toes, fingers or eyes. These slides were filed within the patient's medical records as a visual record of the child and used both to initiate the process of classification and as an ongoing record of their development and to trace the child's features as they change over time:

[The team consider further genetic tests and x-rays of this little boy.]
Consultant: The [family] photos were extremely useful, what we need is a photo for ourselves [to child] your face and hands [SPR takes these photos, front and side of head, hands palm down and feet] first of all has he enough resemblance with J [his father] and M [his mother] that I don't think there's anything significant in his overall appearance to suggest a syndrome. But that doesn't explain his height.

Cataloguing the Rare and Unusual

A number of key features associated with some syndromes are behavioural and within the specialist clinic video recordings of the child was a key aspect of assembling the syndrome, particularly in the context of clinical research. For example, in the case of Rett syndrome, video was routinely used to record the overall 'look' and the behavioural features of the child associated with this syndrome, such as their stereotypic hand movements or their disturbed breathing pattern. However, if the child displayed a particularly florid episode of disturbed breathing (particularly hyperventilation and apnoea) or an interesting or unusual feature of the syndrome, such as a particular level of scoliosis, the team enthusiastically recorded this. This collection captured examples demonstrating the various stages in the

progression of the syndrome and the range of behavioural features associated. It also importantly included examples representing the variability of these features with a focus on the spectacular presentations of extreme or rare variants:

During the history taking, the two consultants look at Tamsin (the same 4 year old little girl) and examined her briefly when she stops and focusses her gaze on the Psychiatrist. They whispered together as they examined her and the Neurologist takes her hand and exclaims "I've never seen that before". She tells the team after the consultation that she was talking about her "shakey limbs" or muscle spasms, which is a rare and unusual feature and they discuss whether this feature is associated with the syndrome.

The team was particularly keen to video Ruby, a rare case, when she attends the clinic with her mother and her grandmother. Ruby has a diagnosis of Rett syndrome but importantly for the team she is considered to be 'high functioning', she has some speech, has the ability to use her hands purposefully and can draw pictures. It is extremely rare to see a girl with Rett syndrome who can speak and communicate. Whilst the consultant geneticist continues taking a history from her mother, the paediatrician (who is a recognized expert in the syndrome) and the SpR take out their small hand held video cameras and start to record Ruby as she sits next to her mum and ask her if she can walk across the room for them. They comment to the room that she has a 'funny flick' of one foot that kicks out when she walks- this is quite pronounced, they continue recording her movements as the rest of the clinical team stop and watch:

Paediatrician: Can you do some things for me first? Ruby, can you do some things for me?

Ruby: Yes.

Paediatrician: Will you stand up for me? Now I just want to see you walking can you walk to the door and walk back?

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Mum: Walk properly you don't have to walk slow – walk properly.

SpR: That's better. She's trying hard not to kick.

Paediatrician: Yes she is isn't she?

SpR: Yes she really is trying there not to go out. A little wobble.

Paediatrician: Very good well done, that's good, right Ruby can you do this one yeah that's it good girl.

The whole team continues to watch Ruby intensely as she walks around the room and the Paediatrician and the SpR continue to video. They are keen to document examples of her speaking, writing her name and drawing for their records and the paediatrician asks Ruby if she can video her while she writes her name and draws a picture of her mother. The paediatrician spends a lot of time working with Ruby, encouraging her to talk to the camera, to draw a picture and to write her name:

Paediatrician: Can you tell me what your name is? Pretend I don't know what your name is. What's your name? Why don't you draw me a picture instead?

Ruby: Yeah.

Paediatrician: You do that. Can you write your name?

Ruby: Yeah.

Paediatrician: You write your name at the top so I know it's yours. OK?

Ruby: OK.

Paediatrician: Good girl. Okay can you draw me a picture of Mummy?

[Ruby starts drawing a picture of her mother- it is a very basic stick figure and the paediatrician moves the camera in close to the child to get the details of her drawing action and the drawing itself.]

Paediatrician: And some arms? And what about some legs? Good girl and now can you write mum, M U M at the bottom just so we know who it is can you do that?

Ruby: Yes.

This is a highly unusual case and is thus highly prized by the team as an addition to their collection of cases and an example of an extremely rare variant within the clinical spectrum of Rett syndrome. Ruby already has the label of Rett syndrome and this is not queried by the team, rather they focus their efforts on recording her abilities (talking and drawing) and the key physical features she displays that are visible behavioral features associated with the syndrome: hand wringing and walking problems. This combination ensures not only that this case is rare, but that others will not query the diagnosis, the visible behavioral features she displays, particularly the hand wringing verify the diagnosis is Rett.

Collecting Cases on the Boundaries

Throughout the course of the clinics the teams consistently show their interest in examining any feature in a patient that may add to their knowledge of the spectrum and variability of a syndrome. A detailed record of interesting cases adds to their database and to their expertise and thus their ability to classify. It also provides the team with the opportunity to test and refine the boundaries of the syndrome and potentially to explore and refine the range of features associated with this entity.

Whilst a small number of boys have been identified with the genetic mutations associated with Rett syndrome, this is an extremely rare variant (such cases are usually associated with extremely severe disabilities and a high mortality rate). Thus, if this boy was found to have a mutation associated with Rett syndrome, this would provide the team with an interesting case. The consultant geneticist describes the association between Rett syndrome and boys and the level of knowledge at present:

Geneticist: Yes, no they did think we did think Rett syndrome was something that just affected girls and in a sense it does, in that, in that, girls with Rett

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syndrome have this sort of seemingly normal development and then lose skills yeah. And the boys who have changes in the same gene don't do, they, they don't seem to have the sort of fine development for a while and then lose skills, so they have....

Mum: To be honest we always said from the beginning....

Geneticist: Yes and we still have, we still really don't know sort of how common Rett syndrome gene problems are in boys. You know we began to get clues, there were a few boys who had, there were one or two boys who were in families with one or two Rett syndrome girls who were found to have changes in the same gene but a much more severe sort of pattern of problems and... died, most of them quite young and so we thought they were very exceptional. But then people have been finding other changes, other changes in that gene that don't lead onto death for boys but lead on to something not too dissimilar to the other syndrome.

More broadly, the identification of cases such as this could potentially contribute to a re-configuration of the boundaries of the syndrome itself – at this time a small number of studies had been published suggesting genetic mutations associated with Rett syndrome had been found more widely in boys with learning disabilities and this case could add to that small body of literature.

Completing the Collection

It was not sufficient to have seen a syndrome, but also to record it. Here the consultant is pleased that this next case provided the opportunity for her to show a patient with a diagnosis of classic Russell Silver syndrome to her junior and a trainee specialist nurse. Not only did this give them a chance to examine a child with this syndrome, but the case notes also held a stack of family photographs. The photographs provide the consultant with the opportunity of talking us through the key physical features of the syndrome, but also to demonstrate the ways in which the features can present differently over time, as the child develops:

[After lunch I go back into the clinic with the consultant and we are all in the room with the SpR, the specialist nurse and a trainee genetic counsellor.]

Consultant: It's a Russell-Silver.

SpR: We've been looking for a Russell-Silver.

[Consultant gets the family photographs from the medical records and we look at them on the desk. They are all photographs of a little boy, one of his birthday party, posing with candles on the cake, one where he is surrounded by hats, others where he is a newborn, a baby, a toddler and later.]

Importantly, despite the child's medical records already holding a large number of family photographs, the team gratefully take another large envelope of photographs from the child's grandmother. The junior (SpR) and the child sit together sorting the pictures into piles according to his age; this is important for the team if they want to capture the way in which the physical features of the syndrome develop within this classic case developed over time.

The Personal Collection: Displaying the Child

An important feature of these visual recordings is that other professionals can see them; patients are presented in absentia and classification can be carried out without the patient present. 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, particularly if a case proves particularly subtle, interesting or difficult to classify. These images are routinely taken to be scrutinised and interrogated for 'clues' elsewhere, most commonly, the local dysmorphology meeting (cf. Shaw et al., 2003), but also at regional, national and international meetings.

Professional Capital

They are also, of course, a form of professional capital and are used for research, journal articles and to enhance expertise and status. These images become part of the clinician's personal collection of types and cases and an important part of the process of developing and estab-

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lishing expertise. In addition, if the case (or syndrome) is particularly 'rare' or interesting, the case can be presented at national or international meetings. In this local dysmorphology meeting one of the consultants is "desperate to show" the group an extremely rare case she has diagnosed; a mother and daughter with Kabuki syndrome. This is a rare syndrome, but it is extremely unusual to have a familial case, both mother and daughter appear to have this syndrome:

Consultant 2: I'm bursting to show these slides....

[We see a series of slides of an 8 year old girl with short brown hair grinning into the camera. This is the case Consultant 2 is very excited about.]

Consultant 2: Would anyone like to make a diagnosis?

[There is silence from the team.]

Consultant 2: This is Abbey, she is doing very well, she's in a regular class, she has some help but is not coping. She came with her mum and dad who want to know what's wrong. She goes on to describe how Abbey was referred by a number of sources "she was also referred by a community health worker I know who suggested I should see her and mentioned that there was something odd about mum too". They go through a large number of slides of the child- head shot, side head shot and details of facial features and hands and feet from various angles, then a slide of the child's mother- head shot of woman in her 30's with short curly hair and large eyes framed by large glasses- the group exclaim then they see this slide.

Consultant 2: She has large eyes, lateral aversion of the eyes [she demonstrates to me by pulling her eyes to the side to get an oriental look], her height is 1.49 centimetres. I got hold of the mother's baby notes, she was seen by lots of paediatricians because of her short stature and her pictures were shown at national dysmorphology meetings in the 70's. So that's mum.

Consultant 1: We're talking about Kabuki aren't we, but the nose isn't.

Consultant 2: The girl does... I'm encouraging mum to get some pictures of her as a child. Mum's got the full house really... mum is so dramatic, I have no doubt in my mind.

They continue to look at slides of the mother and child and discuss that there are no reports in the literature of familial cases of this rare

syndrome. Thus it would be a good case to write up in a journal. This is an important discovery for this consultant, and it displays her expertise in diagnosis, this mother and daughter have been inspected by many other clinical experts who all failed to align these subtle signs. In addition, the rarity of this discovery provides the opportunity to present at a national meeting and to publish. It may also mean that she becomes recognised nationally as the expert in the diagnosis and adjudication of this syndrome.

Personal Witnessing

Clinical consultations also provide the opportunity for the rehearsal of clinical authority. Consultant physicians do not merely display their knowledge of 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 first hand observations. In this clinical team meeting, the consultant starts by looking at photographs that have been sent to her by an international colleague. She makes an instant diagnosis based on the visual signs “all the features are there” and suggests genetic testing to confirm the diagnosis and risk of recurrence:

I arrive at the consultant geneticists office to find a couple of colleagues in her office- a consultant and a specialist nurse. The consultant geneticists has a disk and is looking at pictures on her computer monitor- they are of a terminated foetus. The photographs show a baby with a very large skull, a body shot and the feet. The consultant geneticists notes the toes and very large and splayed in and says “Pifffer syndrome, instant diagnosis,

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dadada! All the features are there, if it's confirmed then the recurrence risk is low. We can do DNA and then the parents".

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 first hand testimony.

Owning syndromes

Individual experts in particular conditions can claim – or be granted – particular privilege in pronouncing on conditions that they ‘own’ by virtue of their special interest, experience, or research activities. Experts such as this are also known by families who usually have a detailed knowledge of the field and the key individuals within it, through patient networks, support groups, and web forums. Parents often demonstrated their expertise and insider knowledge of the field by stating they had met one of the international experts or that a recognised expert in the syndrome had assessed their child. As in this case, the parents attend the clinic with their child who does not have a confirmed diagnosis, but sits at the borders of a syndrome. This claim demonstrates their commitment to obtaining a diagnosis for their child and to their alignment to this particular syndrome:

We actually met (UK expert in Rett syndrome) then. And that was interesting, I wanted to meet her because I'd heard tales of her you know, and we'd filled in a questionnaire for her and sent it off, I wanted to get her opinion.

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 wider dysmorphology team within the department 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 “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 in a general dysmorphology meeting the consultant suggests that they send photographs of the child to a colleague who “is good at spotting Marfan’s”. In the car on the way to a regional clinic we discuss cases and the borderline nature of this case and the conflicting opinions of colleagues nationally and internationally, mean that this consultant is considering sending this patient to the clinician the syndrome is named after:

A potential case of Rubinstein-Taybie syndrome is discussed in a team meeting. The consultant geneticists had sent a suspected case to “a Rubinstein-Taybie guru in Holland”, however the response was that “it wasn’t sufficiently Rubinstein-Taybie”. However, a team member reports that at the London Dysmorphology club “a lot said it was Rubinstein-Taybie”. The consultant geneticists adds “Dr Rubinstein is alive and kicking, so maybe I should send her to him... her face has changed in the last year, but my uncertainty is due to the peer review which is unusual... This conflicting peer review is confusing to me and the parents”.

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.

Collecting, Capturing and Displaying the Child

The clinician’s scrutiny and surveillance of the patient – however cursory or protracted – is endowed with a special significance. The experienced clinician can derive her or his knowledge from a series of sources. The most important is the personal observation of a series of clinical cases. Professional experience, gained through first-hand knowledge, is the touchstone of clinical authority. In practical circumstances of clinical work and talk, such personal knowledge

can take precedence over the pronouncements of other experts, the evidence of current biomedical science, or the formulations of textbooks. Indeed, the personal knowledge of the experienced clinician – as is the case in many other professional contexts – stands opposed to those who might ‘go by the book’. In other words, the experienced physician relies on her or his skill in adjudicating the correct categorisation of abnormality.

This does not imply that the expert clinician has no regard for evidence, nor that the claims of biomedical science are completely overlooked. It would be wrong to suggest that they stand in complete contrast. That is clearly not the case. The clinical expert is a contributor to the clinical literature, and has a thorough working knowledge of the published journal science in the field. But the specifically *clinical* mode of understanding remains at least under-determined by such public evidence. The clinician’s knowledge is not necessarily *tacit*, in the sense that it is often articulated in encounters such as the grand round, the bedside teaching encounter, and the clinico-pathological conference. It is, however, *personal*, in that it is grounded in this practitioner’s embodied practice and her or his biographically-shaped experience.

The contemporary dysmorphology clinic thus encapsulates the epistemological systems of modern medicine, grounded in the clinical gaze and on the classificatory systems of classic nosology. Within such a system of clinical knowledge, the *monstrous* does not escape the boundaries of knowledge. Monstrous appearances, on the contrary, are accommodated and domesticated within the classificatory systems of normal medicine.

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