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Digital imagery and child embodiment in paediatric genetics: Sources and relationships of meaning

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Abstract

Paediatric genetics involves multiple visually based diagnostic processes. While examining the external features of a child plays an important role, of increasing importance are biochemical analyses of blood, which produce digital diagrams that display variations in the shape and composition of chromosomes. The level of magnification and detail that can now be captured is allowing new patterns of variation to be ‘seen’ and possible diagnosis to be made, which were not possible before. However, this generates questions about whether these forms of genetic diagnosis and digital visualisation are increasing the scope of medicine to define the body as ill – regardless of whether symptoms are present. This paper, drawing from research in a paediatric genetic service, cautions against giving too much power to digital imagery. It does so by arguing that the imagery is only one source of visualisation relevant to how the child’s body is read and understood.

Key Words: Genetics, Children, Digital Imagery

Introduction

An important area of paediatric genetics involves establishing connections between problems in a child's development and differences in their chromosomes such as deletions (an area of a chromosome that is missing) and translocations (an area of a chromosome that has become part of another). Some of the associations genetics has identified have become established enough to be given a name, others are so unique they are only referred to via the pattern of chromosomal difference found (for example, '2q37 deletions'). Diagnostic practice involves a combination of physical examination of the child, exploration of family history and cytogenetics - the biochemical analysis of blood samples in order to produce digital diagrams that display chromosomal variation. There have been significant advances in cytogenetics in recent years, which are enabling geneticists to display variations that previously would have been undetected. These include techniques such as Fluorescence In Situ Hybridization (FISH), and Array-Comparative Genomic Hybridization (Array-CGH). Through the use of these new techniques the list of associations that are being made between tiny variations in chromosomes and distinctive patterns of development is growing.

This paper considers whether the new techniques for visualising chromosomal differences provide another example of the power of medicine to define difference and construct disorder. Visualisation is an important tool of medicine and has been vital to its ability to establish authority over other ways of knowing. In cytogenetics the visualisation dynamic is shifting from examining the interior or exterior of the body, towards an analysis of computer produced diagrams that display the affected chromosomes. This paper explores what degree of social significance we should give to this form of visualisation. It does so via an account of the findings from an ethnographic study of a paediatric genetic service where very young children with developmental problems are referred and visualisations – both of the child's physical features and biochemical analyses of their blood – play a part in diagnosis. The paper cautions against giving diagrams too much power by placing visualisations within their broader contexts. Influenced by feminist critiques of medical technology and the sociology of embodiment, the paper stresses the importance of relational dynamics within the production of meaning for both the child's body and the representations of it produced by chromosome diagrams. The paper begins by exploring sociological accounts of visualisation in medicine and of embodiment, before moving on to describe the project and explore the dynamics around visualisation that the fieldwork identified.

Visualising the body in medicine

Medical sociology has traced the evolving importance of visualising the body to diagnostic practice. Drawing from the work of Foucault (1975), it has documented how, from the 18th century onwards, medical training and techniques of reading anatomy for signs of disease have produced a body that has been a site of multiple clues over which the doctor has been 'king' in diagnosis and treatment (Draper 2002). Medical instruments have allowed doctors to cut in to, listen to, and manipulate the human body and freed them from a reliance on patients telling them 'where it hurts'. The medical gaze has made an important contribution to the creation of medicalization - the evolving growth of medicine as the source of explanation for ever wider areas of human life (Conrad and Schneider 1980). Feminists have documented

how women's bodies have been a particular site where medicalization has overwritten other versions of female embodiment (Birke 1999), particularly in areas such as reproduction (Hubbard 1989; Mitchell 2001; Zechmeister 2001). This work has documented how medical technologies enable the clinician to gaze into the woman's body and see things that she cannot (Petchesky 1987; Sandelowski 1994; Mitchell and Georges 1997). For example, Howson argues that 'women's sense and understanding of our "bodily insides" is largely governed by the imagery and discourses of the biological sciences', generating 'a fragmented, objectified and alienated embodiment' (2001: 97).

In contemporary medicine the assumed power of medicalization has been undermined by changes in the organisation of healthcare (Ackroyd 1996; Allen 2000) and patient attitudes (Prior 2003), which have challenged medical authority and countered the medical gaze with alternative ways of perceiving and knowing the body. Nevertheless new forms of visualisation within medicine are still thought to have privileged status, particularly those technologies able to identify illness before it becomes symptomatic (Reventlow et al. 2006; Griffiths et al. 2010). Paediatric genetics is prominent in such accounts because symptoms appear irrelevant to its ability to identify clinically significant variations in genetic make up (Novas and Rose 2000; Featherstone et al. 2005). For example, an editorial in 2008 in the New England Journal of Medicine noted that 'Clinicians, like researchers, can now shift to a 'genotype first' model of diagnosis for children with unexplained developmental abnormalities' (Ledbetter 2008: 1729-30). The implication is that chromosomal differences displayed in computer diagrams are becoming 'the essential referent of new categories of illness' (Navon 2011: 204). Buchbinder and Timmermans argue that you can see this shift occurring in the growing scope of neonatal genetic screening in the US. Here the screening technologies and the molecular disorders being investigated are so new that what is being identified are 'ontologically disputed borderline forms of disease' (2011: 57). For Bleakley and Bligh (2009), influenced by Baudrillard (1994), the use of such technologies symbolises a shift in the operation of medical power, as the ability to produce and interpret a simulation of the body becomes the ground upon which medical power and authority plays out.

Given the explanatory power genetics is often assumed to have, it seems hard to deny the potential significance of visualising DNA. However, there is a danger in arguing that either the classical medicine of anatomy and physiology or the digital world of FISH and Array-CHG are capable of 'producing the disordered body'. Such arguments – like the technologies they study – strip the body of its material and social significance. As Mol (2002: 24) notes '...what is needed, indeed indispensable for clinical diagnosis, is that there be a patient-body'. Neither the anatomist nor the geneticist can go very far without a body and that body does not exist purely in the laboratory or consultation room. It has a life within and outside those spaces where other frameworks of meaning inform how it is read by itself and by others. What this paper argues is that there is a need to explore the interactions between bodies, visualisations and the other materials and actors that participate in the creation of meaning.

To do so the paper draws from both feminist studies of technoscience and the sociology of embodiment. Feminist technoscience studies retains the feminist concern with how medical technologies play a role in confining the possibilities for female

embodiment, however, it does so without seeing such technologies as all powerful (Haraway 1991; Clarke and Fujimura 1992; Braidotti 2002). It acknowledges that innovations in pharmaceuticals (Asberg and Johnson 2009) and neurosciences (Wilson 2004) have the propensity to further advance biomedical versions of embodiment and identity that have the ‘power to define appropriate humanness’ (Asberg and Lum 2009: 333). But it also explores how such technologies can become significant cultural artefacts incorporated into the personhood practices of women and others (Draper 2002; Nishizaka 2011). For example, Roberts argues, in an account of the growth of 4D commercial scanning, that future parents are active in creating meaning from such technology through the ways in which they are ‘mobilising family knowledge and embodied experience in order to narrate the imagery on the screen in a way that is both socially and personally meaningful’ (2012: 299-300).

Contemporary work exploring such networks of meaning are not the first to highlight how bodies are produced through such relationships. Both the sociology of embodiment (Turner 1984; Shilling 1997; Shilling 1999) and symbolic interactionism (Plummer 1995; Maines 1977;) are well established ways of thinking about how bodies are significant, material and relational (Kelly and Field 1996; Vannini et al. 2011). Such work begins in a belief that it is, or should be, impossible to give an account of a ‘self that is foundational without being embodied’ (Shildrick 1999: 78). Debates on embodiment emerged from a disquiet about both classical sociology’s apparent disinterest in bodies (or what Shilling refers to as the body’s ‘absent presence’ in sociology (2003)) and postmodernism’s interest in bodies only as metaphors and symbols (Fraser and Greco, 2005). Various writers such as Foucault (1988), Turner (1984), and Rose (2006) have argued that practices in areas such as medicine, consumerism, and work have heightened the ways in which the body has been incorporated into both expressions of identity and also modes of regulation and social control. In drawing on this work what this paper is interested in is how bodies are not just simply vehicles of external control and manipulation. Instead, the body becomes meaningful through the interactions it is part of, leaving open the possibility of agency as well as regulation (Crossley 2001; Waskul and Vannini 2006; Jackson and Scott 2010). The body is therefore both material and social: ‘simultaneously the site of experience and subject to multiple gazes, inscriptions and positionings’ (Holt 2004: 221). Meaning and selfhood are constructed through our embodied interactions with the social worlds that we inhabit, including the objects, technologies, social contexts and physical spaces that make up those worlds (Blumer 1969; Denzin 1992). What this means is that to understand whether DNA digital diagrams can obtain social significance we have to think of them as only a part of how bodies are defined. What part they will play will depend on the relational networks they are incorporated into.

A focus on diagrams of children’s DNA is a particularly useful way of considering the significance of relationality. All bodies are relational, but in childhood - particularly early childhood - the visibility of the child’s developing body and the role of others in defining it, bring relationality to the fore (Gottlieb 2004; Lee and Motzkau 2011). Brownlie and Sheach Leith propose that ‘babies are positioned at the centre of a network of actors, organizations and material practices’ (2011: 199). The younger the child, the more significant the presence of others in shaping how their body interacts with the world (Mayall 1996) and the more they are read simply as bodies for others to interpret and define. Therefore ‘the infant’s body should be viewed, not as a container for the self, but as the site of *relationality*, the place where the self, others and the world ‘out there’ intermingle’ (original emphasis, Brownlie and Sheach Leith

2011: 202). This is not to say that this is not the case for other bodies (Manning 2009), in particular adult bodies, but rather that the relational practices through which infant embodiment is produced are socially distinctive. In medical contexts technology can become a privileged element in such relationships. Here the child's body is 'constituted by, and contested through, the attachment of non-corporeal, technological elements' (Place 2000: 172). Place's account was based in a paediatric intensive care unit; here the intensity of technological intervention in keeping a child alive and displaying the body's reactions, enable technology to be a privileged actor in the relationships shaping the child's body. In the context of paediatric genetics, the technologies of visualisation exist within a much broader and more diffuse network of relationships that surround the child. What this paper seeks to do is capture how that broader network also provides meaning to the child's body and the significance of that network in defining or refuting disorder.

The paper positions technologies that categorise the body's distinctiveness as one - at times significant - component in relationships of meaning production. Visualisations may be produced in the laboratory, but they live in many different networks, practices, and interactions, which will all play a part in how their meaning take shape, can change, be accepted, rejected or translated. The paper explores both the clinic consultation where visualisations are first displayed and interpreted, and the intimate spaces of everyday family life where such visualisations continue to be interpreted and translated.

Methodology

This article is based on data from fieldwork with 26 families whose young child (under five years old) had been referred to a genetic service in the UK, either during the life time of the project or in the past¹. The service in question is based within one hospital trust, but undertakes clinics across a large rural and urban region. Referrals, usually triggered by paediatricians looking after the child, lead to an initial consultation at one of the clinics. All first consultations are led by a geneticist, although others, including genetic counsellors, paediatricians or medical trainees, can sometimes be present. If, after an initial visual examination of the child and questions directed at the parents regarding family history, the geneticist thinks there is something they can look for, then further consultations and tests occur. Tests do not always directly follow a first consultation, sometimes geneticists wait until the child has developed further in order to establish which chromosomes to explore. During the time period of the project the service was primarily using FISH, although a small number of children did have Array-CGH testing.

Our methodological approach followed each family over time (up to 18 months for those referred to the service during the study), going with them into the different settings of their lives and listening to the perspectives of multiple actors within the family, including parents, siblings, other significant family members and the children marked as different themselves (primarily children who had been referred in the past and who were therefore older than the new referrals). Fieldwork data was generated through a mix of qualitative longitudinal interviews and non-participant observation in clinical and non clinical encounters. Families also shared with us the material they were sent from the genetic service detailing what diagnosis (if any) had been made. In addition, young people and children had the option of filling in journals with their stories, drawings and thoughts on what family and genetics meant to them.

Recruitment occurred through letters of invitation sent via the genetic service (148 in total) and through publicising the research in regional newsletters for families with disabled children. All new referrals to the genetic service of a young child with an unexplained problem in their development over a period of several months received a letter of invitation. In addition a sample (consultants at the clinic did the sampling via reviews of medical records) of families with a child who had previously been seen by the service at least five years before (after checks to confirm the child was alive) also received a letter of invitation. The sample criterion given to the consultants for the past referrals was that we wanted to obtain a balance between those who had obtained a diagnosis and those who had not. One family who came forward to participate were not included in the study, this was because the child who had been referred to the service several years ago did not want to participate and we had a policy of not including families if such children did not want to be involved.

We put in place a number of measures, as part of the overall approach towards protecting the anonymity of the participants, to ensure that the clinic did not know which families went forward to be participants. This included carrying out observations in the clinics with families who agreed to be ‘non-participants’, so that the clinic staff did not know which consultations, and therefore which families, were then included in the analysis and writing up of the data. Given the rare nature of some of the syndromes being examined, information on specific diagnoses is not given here and aspects of the specific variations found have been altered. In addition, information about the families or the clinic that together could potentially identify them is not included. When people shared the letters with us the agreement was that this material would not be directly quoted, but instead could inform our analysis. Therefore, rather than quote the letters we discuss the kinds of material and visualisation found within them. The project obtained ethical approval via the Local Research Ethics Committee (LREC) of the NHS National Research Ethics Service (NRES).

The data analysis within the project involved independent and then shared coding of transcripts and notes within the project team. This process was influenced by a theoretical coding frame that was then applied to the transcripts, observation notes and textual material. The different forms of data were approached as being different in kind and significance. Interviews captured narratives about family members’ lives and the socio-cultural values (or moral repertoires (Ribbens McCarthy et al. 2000)) they used to make sense of those lives (Riessman 1993); the observation notes highlighted the dynamics of social interaction that occurred within specific spaces (Brewer 2000); and the documents produced discursive material that presented a particular way of framing both the diagnosis process and the medical understanding of the child (Fairclough 2001). The value of the different forms of data was how they highlighted contradictions and contrasts within people’s accounts, as well as how they produced an integrated picture. The analysis detailed in the paper is primarily based on transcripts of anonymised interviews with parents, notes of the clinic observations and the clinic letters shared by parents. The observational data enables exploration of the interactions that occurred between the child, the parents, the consultants and the chromosome diagram. The data from the interviews with parents, and their discussion of the clinic documents, enables the parents’ role as socially privileged mediators within the networks defining the shape and meaning of their child’s body to be examined. The patterns discussed below are those that were evident across the transcripts, observation notes, and letters, they are therefore exemplars of the trends in the data.

Displaying the diagram

In all the first consultations that were observed one of the key activities that occurred was the close examination of the physical features of the child. Elsewhere the significance of that process has been explored (McLaughlin and Clavering 2012). In this service such physical examination remains an important starting point for establishing what chromosome variations to look for via cytogenetic analysis. However, in follow up consultations a distinct shift became clear as attention moved away from the child, towards the diagrams that had been produced by FISH or Array-CGH. Below is a typical observational extract highlighting how a geneticist described the significance of FISH analysis:

Geneticist returns to the diagrams... 'So, if we look here [pointing] you can see [numbered chromosome] is in the right place, where the red blob is, [numbered chromosome] is also in the right place, but one has ended up on chromosome [number]. So we had a close look at that. Chromosome [number] had a normal look to it, but here there is a bit of chromosome [number] on [numbered chromosome].'

(Observation Notes, Second Consultation, Rushton Family)

Parents often were impressed by what the diagrams were able to display, the father from the Brown family was amazed by how they could capture such tiny variations within his daughter Grace's DNA:

Ian: He [the consultant] drew a diagram and he said there's like a bar code, you know how they put a bar code on the back of a cornflake box or something? And it's one, it's like one little line and it has like millions and millions of little chromosomesⁱⁱ inside that do different things, and it's one of them that's missing. (Interview Two, Mother and Father, Brown Family).

The Brown family received a diagnosis that their daughter was missing material from a specific chromosome, which had been identified using Array-CGH. In their second interview they discussed how they had been fascinated by this finding and its display for some time - including jokingly calling Grace 'Chromo 12' - for a time. However, their fascination was limited by how little they discovered the hi-tech diagram could actually say about Grace. During the consultation, after going into great detail about how they had been able to produce the diagram, the consultant admitted that due to the newness of the technology there was little they could deduce about Grace's future. Indeed he acknowledged the finding could be a 'red herring' and have little to do with the issues she faced. The Browns found this very frustrating; particularly because they had been called in to hear about findings - for them dots on a diagram were not a finding:

Ian: I think, because we thought we were going to go and they could tell us everything we wanted to know, but they didn't tell us anything, because they didn't know.

...

Kim: in the letter it said, 'when you come for this appointment, as well as the blood test, we'll discuss in detail'

....

Kim: So I thought 'the detail' meant the outlook, but it wasn't, it was the detail about how they did it, how they got to this stage.
(Interview Two, Mother and Father, Brown Family)

This pattern of interaction and disappointment was common as parents tried to find out what the diagrams could meaningfully say about the future for their children:

Geneticist: 'She has a pattern, what we call a Mosaic. This is where people have different pictures in different cells, rather than a single cell being affected. I can't say she won't be having more problems in the future, but what I can say is that, in other cases with similar chromosome patterns, none of them have presented with the same problems. So you should treat her as entirely unique. We will need to keep her under review, to see how she goes'.

Geneticist looks at Lorraine [Mother] and smiles, then looks back down to the page, pointing with a pen, and then up to Jeremy [Father].

Geneticist: 'I'm sorry I can't tell you about what to expect at this stage'.
(Observation Notes, Second Consultation, Rushton Family)

The letters parents received also presented this balance between highlighting the technical capabilities of the cytogenetics analysis via the DNA digital diagram and the uncertainty regarding the long term significance of what was displayed. The letters contained detailed descriptions of what the geneticists saw in the digital images and what they were able to do with the child's DNA. The paragraphs of detailed description highlighted which bits of which chromosome have been deleted or moved onto which part of another, with information about 'short arms', 'translocations', 'deletions' etc. The descriptions were very matter of fact and sometimes were able to name a syndrome, while at other times stayed at the level of indicating that 'der(12)t(12;10)(p7.1 or 2; p15.1)' had been found. However, the letters also contained a great deal of ambiguous language over the future, where the phrases that dominated were ones similar to 'we cannot accurately assess', or 'we cannot accurately predict any problems in the future'. For example, the Hughes family where provided with a named syndrome, but the letter went on to explain that they could not answer the question that mattered most to the mother: what the future held for her son Fred.

For all the authority and solidness of the detail contained in letters, their power is limited to the description of lines and colour; they struggle to translate into knowledge

that is useful to predicting the future or connecting to present or future symptoms. Nevertheless, the letters were often kept in carefully looked after boxes or folders that detailed the child's journey through genetics (and other healthcare providers). It is not unusual for parents of disabled or ill children to meticulously collate and store the medical (and other) documents they receive. These files are a material record of the quest families have been on to find out what lies behind their child's problems. The way they are looked after, stored, displayed and annotated also integrates them into the relational world of the family. Their meaning is produced alongside other pictures of the child and other remembrances of family ties. Therefore, the way the genetic service letters were looked after implies they had a great deal of meaning to the families. Does this mean that the words and the diagrams did come to mediate the parents' relationship to and understanding of their child's body?

The Rushton Family did embed meaning into the letter they received. Lorraine (mother) in particular, had read it many times, trying to make sense of the detailed descriptions she found inside:

Lorraine: we got the letter after the last consultation at genetics, and we can see Alesha's situation is complicated – her chromosome disorder is very complex. It was very difficult to take it all in. I called the geneticist a couple of times after the consultation to ask questions, and went on the internet to try and make sense of what it will all mean for Alesha over time. What the geneticist told us was that Alesha's chromosome disorder is unique to her. She has a balanced translocation, where some of her Chromosome [N] and [N] have swapped over. But also she has a bit of Chromosome [N] missing – what they call a deletion. This is all connected with her medical problems – the difficulties she has are because of her chromosome disorder/translocation. (Second Interview, Mother and Aunt, Rushton Family)

As the discussion carried on, Lorraine picked up the letter and pointed to the diagram showing the chromosome variation and said:

Lorraine: This is Alesha, this is Alesha really, it is her, she is unique. (Second Interview, Mum and Aunt, Rushton Family)

The diagram produced a version of Alesha that was significant to Lorraine and to her relationship to her, but not in a way that objectified and stigmatised Alesha in the straightforward way medicalization would imply. Instead Lorraine drew out a meaning for the diagram that was positive – the language she constantly returned to was of uniqueness – and relational to the other aspects of Alesha that were sustained in her life within the family. While the diagram was an artefact that symbolised Alesha's distinctiveness positively, her mother knew that it could not be a vehicle for defining her now or in the future. This was because all it did was offer one description of one aspect of who Alesha was and her embodiment. It was 'put in its place' by the realities of caring for the actual issues Alesha faced. Making the most of her potential would come via the loving environment the family provided. Alesha was not in the diagram, she was alive, in their home playing. While watching Alesha, with the diagram left to the side, Lorraine went on to say:

Lorraine: What they've said at genetics is they can't say exactly how the chromosome disorder will affect her, because Alesha is unique. But at the moment, most days, I don't even think about it. I just concentrate on getting on with what she needs, and stimulating her as much as I can. [Pause] But then I do think about it – I remember I have a daughter with special needs, she's going to have a different life to what we'd planned.... Genetics have told me as much as they know. So it's a case of like it or lump it, and I just put it at the back of my mind and don't think about it. I have accepted it. And, at the end of the day, she will still have a good life – it will just be different to the one we had expected. But she will still be loved and able to do as much as she is able to.

(Second Interview, Mum and Aunt, Rushton Family)

Once the diagram and text that visualises Alesha's genetic difference entered the relational spaces and connections of family it remained an artefact of display and meaning. However, this occurred through how it was incorporated into the broader versions of Alesha that were maintained by those around her. This wider network – one centred on care – was seeking to find her a place in the world in the future through relationships that recognised difference, but refuted disorder.

The digital diagrams cannot say what the future holds for children like Alesha; only monitoring their development will provide the clues to that. The micro analysis of blood gives one version of what and who the children are, but it is a version that struggles to move past technical description. The diagrams' significance are undermined by their - current - inability to predict future possibilities. As in Roberts' (2012) account of women's consumption of 4D ultrasound, the parents are seeking to give meaning to what the diagrams display, and do so by drawing from broader representations and narratives, which are produced and maintained within familial networks they and their child are part of. In this process differences about their child are not ignored, however their relational framing means that such differences do not over determine how the parents - and others - understand or care for their child. The uncertainties about what the diagram signifies leaves open the possibility that how they care for their child - rather than genetics - will play a role in what the future holds.

Other visualisation practices

The visualisations produced by the geneticists and their labs are not the only visualisations of genetic variation that families incorporate into their approaches to making sense of their child and thinking about their future. It was common (and expected) that families augmented the information they received in consultations with time spent on the internet researching the genetic syndromes and variations explained to them. Parents spoke often about the many websites they visited that mapped the features associated with particular syndromes or variations, and it was clear the images found there were of profound significance to them.

The Todd/Richardson Family were informed via telephone prior to a second consultation that the analysis of their son's FISH tests appeared to show that he had a particular deletion on a numbered chromosome. Soon after the parents received this

information they were on the internet and found images of other children who had what they believed to be the same deletion. They were immediately struck by how similar their child Harry looked to the other children, confirming for them that he must have that deletion. When discussing this with the researcher before the consultation, the father said that when he looked at the images he thought he was looking at his own child. It was therefore a considerable surprise to be told in the consultation that what the geneticists had found was a specific form of deletion pattern on the numbered chromosome that they would not have seen on the internet:

Miranda (mother): 'we've been on the internet and seen lots of things, and some pictures. We can see a lot of Harry in them'.

...

Geneticist starts to explain the chromosome results.

...

Geneticist [pointing to page]: 'You'll see things named this on the internet, with Chromosome [N]. But what Harry has is Chromosome [N] point [N], which is different to what you mostly see on the internet.

...

What Harry has is [pointing to sections of his drawing of chromosomes] one copy of chromosome [N] which is completely normal, and one completely normal but with a tiny bit missing. This can cause a whole series of different things to happen.

....

We see about half a dozen kids a year with chromosome [N point N] deletion and the one thing that is true is that they all vary a bit between them. This is difficult for you, as parents, because we can't say exactly how things will be for each child.'

(Observation notes, Second Consultation, Todd/Richardson Family)

When Bill, the father, reflected later he argued that it didn't matter that his son's deletion was different to those children whose images he had seen:

Bill: 'We could see how all these children look the same. Just having something different about your genes brings them all together, whatever the deletion-point-this-that-or-the-other is'.

(Observation notes, Second Consultation, Todd/Richardson Family)

While the visual analysis of Harry's blood is able to produce significant detail about the specific pattern of deletion, it carries little meaning or value for Bill. As in the other second consultations we observed, the consultant acknowledged it provided little additional knowledge about what the future held. Instead what was meaningful for both Miranda and Bill was seeing other children's bodies whose specific pattern of chromosomal variation was different, but who looked similar. This was emphasised

for the parents when they attended a clinic in London for children with the same or similar variations in their chromosome. In the second interview, Bill described looking at the children in the waiting room as looking at Harry's 'brothers and sisters'.

The Browns had a similar response after going on the internet to research the chromosome variation disclosed by Array-CGH:

Kim: loads of things came up, I mean some were *so* medical you'd just, you know you'd have *no* chance. But I seen this other one that Chromo Twelve dot com and it was from families, it was their own story.

Ian: Yeah, you know, this story that's Grace, that's Grace, Grace does that!

...

Ian: About four different little stories and it was, that's a bit of Grace, that's a bit of Grace, that's a bit of Grace, wasn't it?

(Interview Two, Mother and Father, Brown Family)

For both these sets of parents the visualisation of the specific chromosome deletion did not carry the same level of meaning examining the faces and bodies of other children did. The technical information (*deletion-point-this-that-or-the other*) said so little about Harry or Grace now or in the future, that they could not become a factor in framing them. Nor could they become part of the relational networks that were shaping their children's identity. Instead, networks were being created based on internet images of, or stories about, children who looked similar and actual children met in real life. Again - unsurprisingly in a way - examining images of the external features of bodies carried a greater resonance than what a visualisation - however hi-tech - of minuscule internal variations in chromosomes could do.

Children's bodies, in particular how they look, who they look like and who they don't look like, are integrated into the social and intimate lives of their families (Haimes 2003). In ways that are similar to how Brownlie and Sheach Leith (2011) understand early childhood embodiment, the meanings and connections made to the lived body, within its lived context of family and home, draw from and are woven into social and emotional repertoires of meaning and significance, which reach out and make a connection to other similar looking bodies as equating to a sense of belonging and shared identity. There is a richness to the meaning and value that can be related to the body and other bodies, which the simulation cannot replicate. This is not to suggest that the connection made to the faces of or stories about unknown children found on the internet are more 'authentic'. Rather it is to say that it is easier to make connections and read meaning into the similar physical appearance of different bodies. This is because of the social value given to physical appearance - particularly faces - as part of selfhood and identity (Carsten 2004).

From social anthropology we know that highlighting visual similarities is a form of kinship making, it provides an identity and shape to the child's body, by placing it within the kinship network around it (Carsten 2000; Franklin and McKinnon 2001; Edwards 2005). The kinship making that appeared to occur for the families in the study included drawing connections to others outside the biological family who looked similar to their children. This appeared to offer broader opportunities to

develop relationships and senses of belonging for the child and for the parents. Such kinship making came to the fore in the interviews because of how the genetic diagnosis process was placing those visual similarities or differences within a different relational network – one which sought to identify those physical features with disorder (Fitzgerald 2008). The way that parents and others in the family subsequently talked of their child's physical features, appeared to resist disorder and instead made use of the child's embodiment to retain their presence within the familial network. Or, in the examples where connections were drawn to other children with similar genetic variations, to offer new possibilities for relational connection and belonging. Within all this work producing ties the digital diagram appears relatively unimportant.

Conclusion

The allure of sophisticated visualisation techniques is hard to resist, for clinicians, patients and their carers and for sociological researchers. Caught in the spell of multi-coloured illuminations and graphical displays of the smallest fragments of our DNA there is the potential that we forget to also gaze upon the everyday presence of the actual body in all its messiness and mundane qualities. While the visualisation can only provide an explanation and not support prognosis, parents' investment in it will remain partial. In addition, the visualisation of chromosomes in a diagram is unable to compete with the richness of meaning and relationship already embedded in the living body and its integration into the practises and relationships that inform how it is understood and valued.

The account provided in the paper validates an approach to childhood embodiment and technology, which places both in a relational framework that includes other sources of meaning and identity. In this case the other contributors to shaping embodiment that were discussed were the family networks the children were living within. The recognition of family ties went beyond immediate and biological kin, to also imply a relationship to and connection with children with similar bodies and stories found on the internet or met in hospital waiting rooms. A crucial component of the relational dynamic around the children was one of care, caring for what future could await them and what place in the world could be made for and with them. These relationships were important in shaping the meanings parents and others imparted on their children's developing bodies. They appeared open to an uncertain future, one not locked by framing the body as disordered. Instead the relational meaning given to the children's bodies was one that included a sense that the bodies were different from the norm and often different from their immediate family. However, because the visual gaze associated with the technology could not (nor indeed sought to) impose a reading of the body as pre-determined by its genetic peculiarity, there was a space for relational understandings to form of the body as still to be made and open to a range of possibilities.

Prioritising the relational dynamics involved in giving meaning to DNA diagrams also has implications for debates about whether such technologies leave actual symptoms irrelevant to the identification of illness and disorder. The account given here implies that symptoms can continue to matter. The diagrams did identify variations that took on some meaning for parents, but while the correlation to current symptoms was weak and while their ability to say anything about future symptoms was minimal, the space

remained for the parents to approach their children's bodies as not pre-determined by the lines and dots of the diagram. What remains to be seen however, is whether advances in what genetic technologies are able to - or claim to - predict about the future might change that. Much research is being invested in Array-CGH and even yet more sophisticated visualisation techniques, such as whole exome sequencing. An important aim of this research is to enable the visualisations to become more powerful in prognosis as well as diagnosis. The hope is that large enough population studies of groups linked to conditions such as Autistic Spectrum Disorder (Pinto et al. 2010; Shen et al. 2010), will allow geneticists to disentangle genetics from all the other social and environmental factors that influence such conditions. If such a goal is achieved (which is a big if disputed by geneticists as much as anyone else) then some of the power being proposed for such simulations of the body may begin to appear. The spectacular display of minute variations in genetic material then may take on the ability to define body disorder without the need for the body itself to display and enact symptoms of difference. However, while such techniques remain at the level of description and explanation, the way they interact with the relational bodies who's DNA they display will limit their ability to define and produce disorder.

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ⁱⁱ He means genes here

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Biography

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