Genome Presence:
The Work of a Diagnostic/Iconic Image

Aryn E. Martin

Supervisor: Joan Steigerwald

Volume 6, Number 5
FES Outstanding Graduate Student Paper Series
May 2002

ISSN 1702-3548 (online)
ISSN 1702-3521 (print)

Faculty of Environmental Studies
York University
Toronto, Ontario
M3J 1P3

Abstract

This paper is an exploration the work of a potent image: the human-instrumental-material work required to produce a karyotype, as well as the work done by the image, both in clinical settings, and as a public emblem of "the human genome". In keeping with theoretical accounts of the visual in science studies, I conduct an ethnographic exploration of the resolution of ambiguous bodies into their genomic portrait. Next, I leave the specific context of the image's production to speculate on the existence of "genome presence", which, much like "fetal presence", relies on public consumption of newly visible objects.

Introduction

> [M]icroscopic objects, particularly chromosomes, are themselves objects of beauty, analogous to the beauty of a Rembrandt… [C]hromosomes have attracted many microscopists not only because these sausage-like bodies represent vehicles of genetic material (and, hence, are biologically important) but also because they are hypnotically beautiful objects. (Hsu 1979, 4-5)
> *T.C. Hsu, renowned cytogeneticist, Human and Mammalian Cytogenetics, 1979*

Perhaps chromosomes are biologically important because they are beautiful. Or perhaps the beautiful and the biologically "real" merge together seamlessly, so that we cannot say whether a karyotype - the portrait of chromosomal material in a cell - is aesthetically beautiful *or* clinically informative (see figure 1). Instead, its meaning is accomplished by overlapping and mutually constitutive features - material, semiotic, aesthetic and scientific. The karyotype is a symbolic pictorial arrangement of human chromosomes, 23 pairs in a normal cell, which stands in for the entire human genetic complement of a body. Each chromosome is a multiply twisted and condensed piece of DNA, housing hundreds of genes. If we envision cytogenetic practice as a chain of translations from bodies to laboratories to diagnoses, the ordering of chromosomes into a karyotype is a salient moment of stabilization. Preparatory laboratory procedures of collecting cells, isolating, staining, fixing, and viewing through a microscope all point to this moment of visual recognition. The diagnosis, counselling, decision to abort an abnormal fetus; all this follows from it. The image itself is the temporary stabilization of an otherwise messy melange of biological, political and practical contingencies. When the karyotype image is taken to be a gaze at the genetic portrait of a prenatal body, the work of "making visible" is effaced, and the genome appears to be a thing-in-itself.

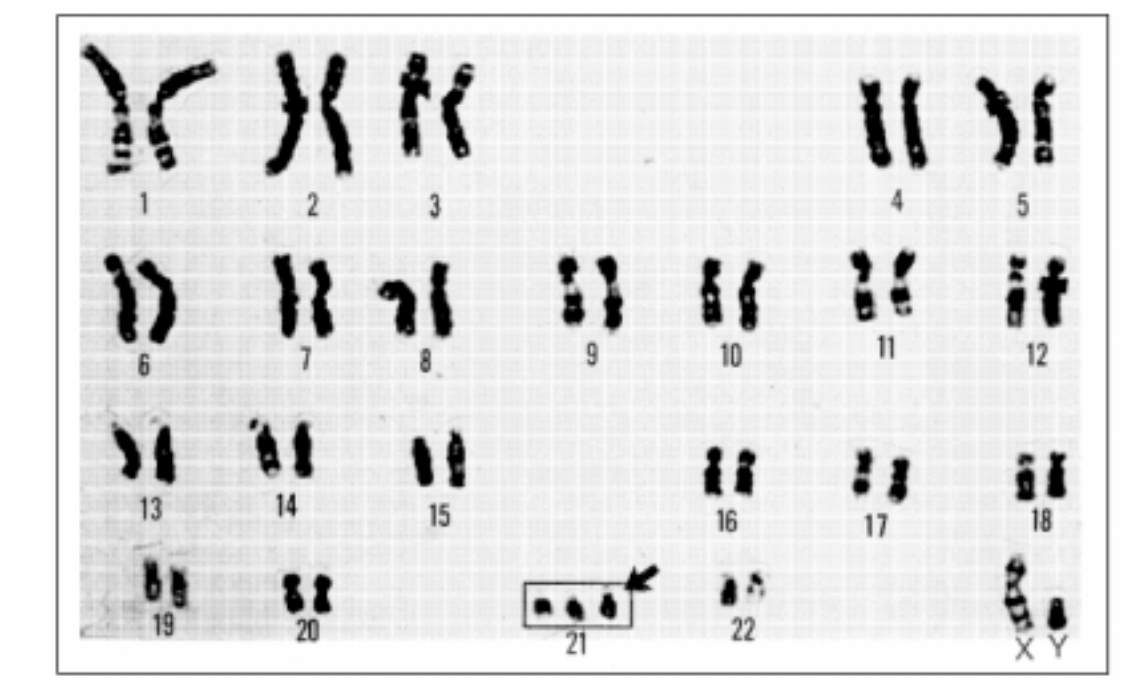

Fig. 6. Karyotype. *Microscopic examination of chromosome size and banding patterns allows medical laboratories to identify and arrange each of the 24 different chromosomes (22 pairs of autosomes and one pair of sex chromosomes) into a karyotype, which then serves as a tool in the diagnosis of genetic diseases. The extra copy of chromosome 21 in this karyotype identifies this individual as having Down's syndrome.*

Figure 1: A sample karyotype, taken from the Human Genome Program's *Primer on Molecular Genetics* (1992)

An alternative reading of this image is that cytogeneticists and technicians are engaged in collective aesthetic projects such as recognition, coherence, fragmentation, abstraction and mimicry. Intelligibility, competence, and communicability are rendered possible by a chain of social and visual conventions, historically established and enforced by standards of cytogenetic nomenclature (ISCN 1995). With Michael Lynch and Samuel Edgerton, I argue that "[t]his hands-on process of interpretation can be treated as an art situated within the performance of scientific practice" (1988, 212). My argument stems from a conviction that visual coherence and pleasure is not an incidental or trivial aspect of chromosomal imaging and diagnostics, and that the resultant image - a karyotype - achieves a stability which belies its contingent

underpinnings which can be located in the work practices of laboratory cytogenetics. Moreover, the image itself accomplishes material effects in the world.

In what follows, I will explore the work of an image: the human-instrumental-material work required to produce a karyotype, as well as the work done by the image, both in clinical settings, and as a public emblem of "the human genome". As this paper is a condensed account of a larger project, it has some methodological disjunctions, which I hope will nonetheless hang together in a coherent story. First, I will introduce theoretical approaches to the visual in science studies, a literature in which my project is situated. Second, in the spirit of laboratory studies[1], is an ethnographic exploration of the crafting of a karyotype. This account of the minute details of turning a body into an image is based on my own experience in a cytogenetics laboratory (as an undergraduate in 1995-96), and subsequent research in a variety of labs, where I observed practitioners in their mundane activities of karyotype-making. Finally, in keeping with works in feminist cultural studies[2], I leave the specific context of the image's production to speculate on the existence of "genome presence", which, much like "fetal presence", relies on public consumption of newly visible objects.

'Seeing' in Science Studies

Much recent work in science and technology studies has interrogated the role of inscriptions and visual images in scientific practice.[3] The present study will draw upon these resources, and contribute to the accretion of case studies in the field. Despite growing attention to visual practices, we do not yet have a coherent body of work "not only because the term visual representations covers a wide array of devices and practices but, more important, because of the diverging approaches brought to bear on the issue" (Cambrosio et al 1993, 663). The inscriptions with which various authors engage include photographs, sketches, diagrams, cartoons, graphs, autoradiographs, etc. Moreover, attention has been paid to diverse aspects of visual inscriptions,

[1] Classic lab studies include Laboratory Life by Bruno Latour and Steve Woolgar, work of Michael Lynch, and of Karin Knorr-Cetina.
[2] Such as Donna Haraway (1997), Lauren Berlant (1997), and Barbara Duden (1993)
[3] Lynch (1985, 1988, 1990a, 1990b), Latour (1990), Cambrosio et al (1993), Fyfe & Law (1988), Knorr-Cetina & Amman(1990a, 1990b), Ruse and Taylor eds. (1991)

including the work practices involved in the production of images in laboratories (Knorr-Cetina and Amann 1990a, 1990b; Lynch 1988, 1985), the serial translations through which images are transformed (Lynch and Woolgar 1990; Latour 1995), the political and polemical role of images (Latour 1990; Cambrosio et al 1993; Fyfe & Law 1988), and the semiotic gestures of images published in scientific articles (Bastide 1990; Lynch 1985).

Observation is foundational to science, and the images created through visual technologies -human and instrumental- have, since the Scientific Revolution, been imputed with epistemological significance; they have been likened to the "language of the phenomena themselves" (Marey, from Daston & Galison 1992). Experimental observation, and the images used to convey knowledge of nature in published scientific accounts rely on this deeply embedded notion that "the process of looking is central to the acquisition of valid knowledge of nature" (Jordanova 1989, 91). Barad notes this historical assumption that takes "observation to be the benign facilitator of discovery, a transparent and undistorting lens passively gazing at the world" (1998, 7). Haraway's ironic use of the phrase the “view from nowhere” plays upon the hegemonic practices of visualization, where unmediated truth is accessed through an "ideology of direct, devouring, generative, and unrestricted vision" (1991, 189). Hence, vision, or more precisely the act of looking, is a powerful metaphor for the acquisition of scientific knowledge. "In the natural sciences evidence appears to be embodied in visibility; in a literal sense, it is embodied in what we can see on a data display...Among [the] modes of relating to an object, only seeing bestows on objects an accent of truth" (Knorr-Cetina & Amman 1990a, 86).

Recent science studies scholarship has problematized any inkling of an unmediated gaze upon nature. "Visibility is not transparency. Rather... visibility is itself a claim that must be carefully examined: in acknowledging what is seen, and newly seen, we need to be equally vigilant about what is not seen or no longer seen" (Treichler, Cartwright & Penley 1998, 3). We know objects only through representations (Lynch & Woolgar 1990; Rheinberger 1998; Hayles 1993). However, "there is no such thing as a simple representation of a scientific object in the sense of an adequation or approximation of something out there, either conceptually or materially" (Rheinberger

1998, 104). This position denies the possibility of a copy theory of visual representation, because the "object" can only be another representation. Hence the pictures scientists make are not a window to nature but crafted representations which gesture toward an unknowable referent. Although at least part artifice, they are necessary in order to interact with the world in meaningful ways; representations are all we have to work with. In their introduction to Representation in scientific practice, Michael Lynch and Steve Woolgar (1990) contemplate what a sociological analysis of the "contents" of scientific knowledge might entail. Although diverse and heterogeneous, inscriptions are an important resource in scientific fact-making, and hence a critical site for sociological analysis:

> Manifestly, what scientists laboriously piece together, pick up in their hands, measure, show to one another, argue about, and circulate to others in their communities are not "natural objects" independent of cultural processes and literary forms. They are extracts, "tissue cultures," and residues impressed within graphic matrices; ordered, shaped and filtered samples; carefully aligned photographic traces and chart recordings; and verbal accounts. These are proximal things taken into the laboratory and circulated in print, and they are a rich repository of "social" actions (5).

Further, Lynch's (1993) description of the "externalized retina" is useful in considering the making of scientific images as a social process. Lynch displaces the idea that image perception occurs in the individual's mind with an emphasis that the selection and circulation of images is a social activity, which involves a great deal of disciplinary convention. The crafters of image in science have predispositions about what something should look like, which shapes what the object is made to look like. The resulting representation, then, makes the conventions all the more robust.[4]

[4] Lynch and Edgerton describe this process with regard to astronomers' visual practice, 1988.

One aspect of the sociality of vision in science is aesthetic convention. Aesthetic criteria are paramount in the determination of which images most accurately and most convincingly represent nature (Lynch & Edgerton 1988). Pleasure, I argue, is a potent motivator of the skilful manipulation of experimental conditions, the endpoint of which is a pleasing image. Pleasure and aesthetic reward is also a culturally and historically contingent variable; the tacit criteria for a "convincing" or "beautiful" image in scientific testimony intertwine with established disciplinary and social conventions. The rewarding product of visual tinkering, in contemporary scientific image making, is an image whose boundaries are well defined, colour enhanced and constitute a clearer representation of nature than the messy materials with which the inquiry began. The irony, of course, is that the greater the degree of image-processing, and the more thoroughly the hand of the scientist/artist/instrument is implicated, the more likely we are to believe that what we are seeing is "real". Recognition of a scientific object goes hand in hand with aesthetic reward, all the while obscuring the cascade of abstraction from the original material.

Tailoring pictures and "cleaning the data" are practices imbued with aesthetic criteria, where representational realism is often the goal. There is an intimate association, Lynch and Edgerton propose, "between [contemporary] science and a particular version of aesthetics emphasizing the simplicity, 'graphical elegance', and representational utility of compositional detail" (1988, 213). Technoscientific apparatuses of visualization and image production allow almost infinite possibilities for poking and prodding, enhancing some characteristics while obscuring others. That these modifications are governed by implicit and explicit aesthetic criteria implies a layering of social intervention in the constitution of a scientific fact. That they are contingent upon historical and cultural conventions and preferences, suggests that aesthetic criteria constitute an element of scientific fact-making.

The Crafting of a Karyotype: Translation Work

Keeping in mind the above theoretical considerations of scientific image-making, I will now examine a very specific ethnographic account of the production of a karyotype. The purpose of this micro-analysis is to open or to destabilize the image; rather than a

thing-in-itself, the karyotype is a labour-intensive accomplishment. In his intriguing "Photo-Philosophical Montage", Bruno Latour (1995) follows a field trip wherein a group of researchers transforms the forest of Boa Vista into a diagram, through a series of intermediary translations.

> Is the diagram…more abstract or more concrete than at our previous stages? More abstract, since here an infinitesimal fraction of the original situation is preserved; more concrete since we can grasp it in our hands, and see with our eyes, the essence of the forest transition, summarized in a few lines. Is the diagram a construction, a discovery, an invention, or a convention? All four, as always. The diagram is *constructed* by the labors of five people and by passing through successive geometrical constructions. We are well aware that we have *invented* it and that, without us and the pedologists, it would never have appeared. Still, it *discovers* a form that until now has been hidden but that we retrospectively feel has an eternal presence. At the same time, we know that without the *conventional* coding of judgements, forms, tags, and words, all we could see in this diagram drawn from the earth would be formless scribbles (177-178, original emphasis).

Like the diagram in Latour's study, a karyotype is constructed, discovered, invented and conventional. A karyotype is the result of a work process - progressive alignments of representations. It is a stabilized inscription that, with the help of conventional dictates of interpretation, speaks for the body. Yet it is also "loaded with reality" (Latour 1995, 175). A karyotype showing three copies of chromosome 21 (figure 1) has some "real" correlate with the physiology, appearance and behaviour of the Down's syndrome body which carries the cells from which the picture was abstracted. The reality of the diagnosis, however, is neither there-in-the-body, nor externally imposed upon it, but emerges with the pictorial karyotype.

Michael Lynch (1985) observes that with normalized graphic display, the rat as laboratory specimen becomes both more than, and less than a rat. Similarly, the body that has been marked by a karyotype, and diagnosed as abnormal, is *more than* the

legions of unsuspecting bodies whose chromosomes have never been extracted, stained and ordered. Reducing a body to its chromosomal content also makes it *less than* the person - with personality, desire, goals, flesh, bones etc. - from which it came. In prenatal diagnosis, karyotyping often renders the body disposable through selective abortion. With attention to translations and serial chains of representation introduced by Latour, I will proceed to locate the karyotype in such a chain.

The bodily source of the tissue to be examined appears at the beginning of the series. Patients of cytogenetic testing are often "high risk" pregnant women, newborns and toddlers with undiagnosed abnormalities, and infertile couples who wish to discover potential genetic reasons for their inability to conceive. Increasingly, chromosomal screening is being applied to embryos prior to implantation, in an attempt to circumvent pregnancy with an "affected" fetus. These patients are of course embedded in heterogeneous networks of political, moral, biomedical and cultural contexts. An attempt to unpack the ambiguities in the lives of patients is beyond the scope of this paper, although not entirely irrelevant to my broader project. A particularly contingent and relevant consideration may be: who finds themselves being tested in the first place? Access, geography, economics, directive counselling, biomedical vs. anti-biomedical orientations, etc. all play a role in determining whose cells end up under the microscope.

Once enrolled in the procedure, patients undergo sample collection. While blood is fairly easy to collect, amniotic fluid collection from pregnant women is highly invasive, risky and often needs to be repeated in order to obtain viable cells.[5] Moreover, preparation of metaphase spreads (the only time chromosomes are visible as individual fragments) requires growing the cells in culture - a finicky procedure with variable success. The fundamental premise of chromosomal structure is that the chromosomal complement of every cell is the same, so visualizing a blood cell characterizes every cell in the body, which enables us, in principle, to specify "a karyotype" for each individual. The ever-present threat to this premise is that each patient could be a

[5] For a thorough account of amnioscentesis, see Rayna Rapp's Testing the Woman, Testing the Fetus , 1999. The book includes a chapter on laboratory cytogenetics, published since this paper was written.

mosaic, indicating that they have more than one cell line.[6] Mosaics can have two or more distinct karyotypes. The ambiguity that this introduces at this stage in the chain is that ideally cells should be inspected from several cultured cell lines. Limitations of material practice sometimes make this impossible. Moreover, the test will necessarily be limited to the tissue type collected, while the patient could be mosaic in other tissues. Hence, the cells which end up under the microscope slide may not be representative of an entire body - an ambiguity often hidden when a karyotype is offered as conclusive.

The cascade of fixation from bodily material to prepared slides is a laboratory performance involving arresting the cells at metaphase by the addition of colchicine to "shorten the chromosomes, destroy the mitotic spindle, and as a result spread the chromosomes around the cell" (Therman and Susman 1993, 33). Cells are treated with hypotonic solutions to prevent chromosome clumping and unfetter the chromosomes of membranes, proteins and cellular "debris" which inhibits clear viewing of chromosomes. Next cells are chemically "fixed" to preserve their morphology, and dropped or sprayed onto microscope slides. Once on the slides, chromosomes are "banded". Staining chromosomes is akin to labelling, or writing on the chromosomes to differentiate chromosomal material so that it becomes visually distinguishable. "The stain, like a name, defined, pointed to and consolidated the visibility of its object" (Lynch 1985, 51). In the series of translations, microscope slides are closer to a two-dimensional surface on which we can find meaning which stands in for the body and the messy tissue from which it came. They are also hybrids of nature and artifice. Something of the body exists on the slide, but it has been killed, prodded and transformed by human intervention.

Two relevant observations can be made about this step (which takes several days) in the transformation from messy bodies to microscopically analyzable display. First, the teleology of each of these translation practices is to allow, enhance and achieve *visibility*. "Such displays systematically transform specimen materials into observable and mathematically analyzable material…the artificial appearance of a specimen is what enables it to be observed in the first place" (Lynch 1985, 37-38). In

[6] This is a well recognized phenomenon in cytogenetics, and has been described to me in interviews as an ever-present concern in prenatal testing. The case in which I was involved, described in Khalifa et al,

the series of translations, the laboratory regimen is designed to yield the one extractable piece of desired information: the visible karyotype.

The second observation is that considerable tinkering and ritual is involved at each stage of preparatory laboratory work. Like the plasmid prep in Jordan and Lynch's ethnography, the "metaphase spread" is achieved through almost imperceptible variations in each lab, each time a slide is made. "While practitioners are assured that the plasmid prep is simple and routine, and while they accomplish it habitually, occasional questions and disputes arise as to just what that routine involves and just how ritualistic its performance should be" (1992, 106). An anecdotal variance I have observed in cytogenetic practice is the distance between dropper and slide which can vary from several inches to several feet - ostensibly to summon gravity to help with cell flattening. When a researcher climbs on a table to aim a dropper toward a slide on the floor, preference, superstition, technical competence, aim and performance art are seamlessly intertwined. Part of the visual reward, the beauty, when one finally looks through the microscope is the realization that this game of technique and ritual yielded anything intelligible at all, much less a spread that will photograph nicely. The cytogeneticist, in this sense, is like an artist who paints blindly for days but cannot see her canvas until she has put away her palette and paints.

Microscopy is the unveiling moment in this succession of translations, wherein bodies become blood becomes cells becomes chromosomes on a microscope slide becomes karyotype. Sequestered in a darkroom, with an elaborate and expensive microscope, a cytogeneticist or technician endeavours to make sense of what was before elusive and hidden. A karyotype is constructed, discovered, invented and adapted to visual conventions. A photogenic metaphase spread is extracted from many potential candidates. A slide will contain hundreds if not thousands of cells. Most will be overlapping, inadequately fixed, stained or preserved, and encased in cellular gunk. If the ritual of pretreatment was effective, however, some will be sufficiently spread apart and unfettered of cellular contents to allow individuation and ordering of the chromosomes. The reward in cytogenetic tinkering is a cell in which the chromosomes are well spread, clearly banded, not too long or too short, and lying in one plane. When

was an incidence of mosaicism.

Dr. Hsu accidentally discovered hypotonic pretreatment in 1952, he described it in these aesthetic terms:

> I could not believe my eyes when I saw some beautifully scattered chromosomes in these cells. I did not tell anyone, took a walk around the building, went to the coffee shop, and then returned to the lab. The beautiful chromosomes…were still there; I knew they were real (1979, 17).

As Ian Hacking describes in Representing and Intervening, seeing through a microscope is achieved "by doing, not just by looking" (1983, 189). Cytogeneticists skilled in their craft have seen countless metaphase spreads. They are intimately embedded in the tiny world to which they exclusively attend, scrolling around in a spread, moving the plane of vision through all three dimensions, distinguishing a chromosome from overlapping bits that occlude recognition. "The microscopist moves around in microscopic space the way a human body moves through a space bodily surrounding him or her" (Rasmussen 1997, 239). Nicholas Rasmussen, in describing electron microscopy suggests that the instrument mediating vision "becomes so familiar it effectively disappears and becomes an organic extension of the user's sense organs and limbs" (228).

What are the visual and semiotic conventions of establishing order and coherence in this particular display? Several authors before me have considered the iconography of scientific image, and my analysis draws from their work (Knorr-Cetina and Amman 1990a, 1990b; Lynch 1985; Bastide 1990; Cambrosio et al 1993). First, we must notice that the series of translations has moved us from microscopic space, which is three dimensional, if only in a very thin context. A two dimensional photograph of the spread, captured by specialized computer software, allows us to physically cut out and arrange the chromosomes in the medium of a printed page.[7] As Bruno Latour observes, "Yes, scientists master the world, but only if it comes to them in the form of two-dimensional, superimposable, combinable inscriptions" (Latour 1995, 147). The first obstacle is overlapping chromosomes, inevitably found even in the most beautiful of

spreads. This is overcome by taking more than one picture of the spread, so that an intact copy of each can be extracted. Once cut out, or selected by a click of the mouse in more sophisticated imaging software, they are moved about into a pattern dictated by the International System for Human Cytogenetic Nomenclature (ISCN). This pattern was arbitrarily decided at the Denver conference in 1960, based on chromosomal length. It was revised in 1971 at the Paris Conference, where some reshuffling took place due to improvements in technique, the most significant of which was banding.[8]

There is a "right" way to make a karyotype. Chromosomes are aligned from largest to smallest, and grouped according to the location of their centromere (constriction). There are four layers or levels, and the maximum number of pairs across the page is seven, in the second level, if counted from the top. They are oriented vertically as if standing up, with the shorter arm (the "p" for petite arm) pointing to the top of the page. Several assumptions guide a karyotypist. First, chromosomes usually come in pairs, one of maternal origin and one of paternal. Second, each pair of chromosomes soaks up stain predictably, so that "bands", or transverse regions of light and dark, are the same within a pair, and distinguishable from other pairs. "The banding patterns of each pair are unique and exploit an underlying variation and DNA structure and activity in different parts of the chromosomes" (Rooney et al 1992, 5). Predictable banding allows us to match analogous chromosomes and compare their banding pattern to the photographs and ideograms made available in the ISCN. Part of the artistry of the practice is learning to distinguish among chromosomes. Rayna Rapp (1999, 203) describes some of the tacit rules of recognition:

> Techs taught me to identify chromosomes by personifications alleged to be carried in the contrastive bands: the 7s, 8s and 9s are deemed to have distinctive and recognizable noses, glasses and beards; 12s, 13s and 14s sport heads, shoulders, and hips; the X-Y sex chromosomes are stereotypically identified by bikinis and chests, occasionally characterized as "wimpy."

[7] When I speak of "the chromosomes" here, I mean a picture of them; their reality is sanctioned by the representation in hand, and their materiality is understood as inextricable from the representation (Lynch and Woolgar, 1990; Cambrosio et al, 1993).

[8] "Banding" refers to staining the chromosomal material so that each chromosome has a characteristic profile of dark and light regions.

This delightfully descriptive language suggests that convention building is more than just visual, and operates through "shop-talk" as well. The rule of uniform banding has infinite exceptions, as there is variability among even "normal" individuals (some "band nicely"), and with adjustments in pretreatment procedure (Rooney and Czepulkowski 1992).

Banding is an interesting site for examination of the historical stability of conventions in cytogenetics, and the practical contingencies that allow artificial intervention to become built into a scientific object. This is a case where "the artificial appearance of a specimen is what enables it to be observed and analyzed in the first place" (Lynch 1985, 38). Chromosomal nomenclature is parasitic upon banding patterns, and banding with particular stains (commonly quinacrine or giemsa) has become reified by standardization. Not only are chromosomes "named" (numerically), communicability relies on standardized numbering of "regions" within each chromosome, which are divided by "landmarks", or obvious bands.[9] The numbering of regions begins at the centromere and moves out in either direction. An interesting anachronism of this naming convention is that with increasing resolution power, bands are being subdivided into smaller and smaller increments, each subdivision limited by 9 digits. For example, 1q24.3 means chromosome 1, long arm, region 2, band 4, sub-band 3. For a more clear description of nomenclature, refer to ISCN (1995) (pictures really are needed to do justice to this discussion!) Suffice it to say that the materiality of chromosomes is embedded in, and inextricable from, conventions of staining and naming.

Once ordered, the unruly chromosomes have become tame, or in Lynch's terms, docile. "A docile object is the product of [marking, constituting graphic space, and normalizing observation]. It is an object that 'behaves' in accordance with a programme of normalization" (1985, 43). This selective accomplishment of interpretative convergence through graphic display is a theme common to analysis of visibility in science studies. Francoise Bastide (1990) suggests that framing and focusing result in

[9] The terminology - "landmarks" and "regions" begs a mapping analysis beyond the scope of this paper.

channelling of meaning in a pictorial presentation. Framing is the reduction of noise, and focusing "consists of cleaning away the nonpertinent elements from the image" (207). Karen Knorr-Cetina and Klaus Amman focus on autoradiographs to describe the processes of fixation which turn data (e.g. the metaphase spread) into evidence (e.g. the completed karyotype) (1990a). Objects, they argue, are visually flexible phenomena, whose definitional boundaries are at stake. "Data become evidence only after they have undergone elaborate processes of selection and transformation" (88).

What kind of evidence is a karyotype? This two-dimensional pictorial convention is a pedagogical enactment through which students learn their craft. Furthermore, it is routinely performed for the purpose of reporting; the karyotype is a reassurance or guarantee of what was seen. It is passed from the technician to the lab director, who "signs off" on the diagnosis, and it is captured in an archive. Its currency is established when it is shared with the presiding doctor, the patient or parents, scholarly communities through publication, and the courts if the diagnosis ends up in dispute. Its stability is reified as it passes through successive hands. Rayna Rapp highlights cytogenetic practitioners' awareness of the work of producing a visible diagnosis: "Laboratory workers tolerate a high degree of ambiguity in the process of reaching consensus diagnoses. But once that process is complete, they must also accept the closure of disambiguation" (1999, 205). This closure is embodied in the visible karyotype.

Material Effects: the Career of a Diagnostic Image

The remainder of the paper is concerned with the material, or "real life" effects of the karyotype image, post-construction. I will make a distinction here between the clinical karyotype, and the "iconic" karyotype. The former has a specific medical context, and is attached to a particular body, while the latter has been detached from its corporeal specificity and functions as an exemplar, much like "The Human Genome" which doesn't belong to anyone in particular, but is said to implicate each of us. In either case, my contention is that the image has instrumental effects that range from terminating a "marked" fetus to infiltrating experiences of pregnancy, reproduction and selfhood.

In a clinical setting, the diagnosis is dutifully recorded in a report, with two or

more aligned karyotype photographs to act as visible and legal guarantors. If the karyotype was normal - 46 intact chromosomes - the chain ends here, or diverges into other tests, if a clinical pathology remains undiagnosed. The disorder may still be genetic in origin, but not at a level visible at cytogenetic resolution. If the test was prenatal, the mother will likely continue her pregnancy happily, reassured by the biomedical stamp of approval. Many women describe that they consider their pregnancy tentative until cytogenetic testing is complete. The stability of the image belies that this reassurance is somewhat spurious, given the range of possible problems, genetic and otherwise, which the test neither seeks nor finds.

If the test revealed a deviance from the expected norm, the body from which the cells were extracted becomes "marked" by difference. This can have a variety of outcomes, including termination of a pregnancy (the only "treatment" available for most anomalies), or assignation to a "syndrome" group which labels and to some extent influences the life path of the individual. Often the information is largely useless to the patient (as in the case of a first report of a chromosomal abnormality), but becomes a publication in a journal of clinical genetics.[10] "47, XY, +21" is the summation of a picture (figure 1, for example). It is an equation, a symbol, a stand-in for the body-cell-slide-photograph from which it came. Its simplicity, however, is misleading. Families receive, interpret and internalize genetic information in diverse ways.[11] Moreover, the simplicity implied by the equation is muddied when we consider that it doesn't predict the severity of expression of genetic abnormality. While ambiguity is temporarily resolved, and coherence achieved by a diagnosis, messiness continues to flourish when the information obtained leaves the hospital.

The "iconic karyotype", as I have termed it, is like the image depicted in figure 1, taken from the US Department of Energy's "Primer on Molecular Genetics". As opposed to the images encountered in diagnostic settings (probably the uncited origin of this particular photograph), these public-sphere reproductions of the karyotype are doing a different kind of work. My premise is that images can provide us with a powerful

[10] The project in which I was involved falls into this category. It was the first reported case of monosomy 18 mosaicism (Khalifa et al, 1996).
[11] Rapp, 1999, does an excellent job exploring some of the complexities faced by women undergoing genetic testing.

site for examining sites of struggle and meaning-making. They are persuasive elements in public understanding of science. Particular images gain credence through repetition, promulgation in arenas apart from their laboratory source, and when they are coupled with scientific authority. In these functions, images act as tools of fixation, establishing in our memories a trace, with which we associate the determinative power of genes. However, because images are subject to diverse readings, are always already impacted by prior tacit knowledge of the observers, and have a life of their own beyond the intent of the producers, fixation is never wholly successful. Like metaphor, visual imagery can be understood as a creative and contingent mediator in the communication of scientific "fact".

Treichler, Cartwright and Penley (1998) have recently edited a collection, The Visible Woman, which places visualization at the centre of scientific, medical and cultural understandings of health and the body. They assert that "given the power of these new [visual] technologies and their claims of unprecedented access to the natural world, it is not surprising that we acknowledge and think about the nation's health crises - whether biological, social or environmental - through the visual images most readily available" (2). Pictures give us tools to think with and ways to understand complex problems. Treichler et al suggest HIV as a virtual "superstar" whose schematic diagrams and 3D models have helped us understand the rudiments of how the virus works. "Well acquainted with the conventions of scientific representation these viral images embody, we recognize them as what viruses are supposed to look like; the idea and the image have come to seem natural to us" (2). However the images that shape cultural understandings of science are neither innocent portrayals of nature, nor are they imbibed unproblematically and uniformly by diverse audiences. "Hence whatever our practical, personal responses to and experiences of these sophisticated new imaging technologies, it is crucial that we understand their performative character, that is, their role as a staging ground for struggles over agency and control" (Treichler et al 1998, 5).

In Picturing Power, Fyfe and Law (1988) similarly argue that pictures are implicated in questions of power and knowledge:

> A depiction is never just an illustration. It is the material representation, the apparently stabilised product of social difference. To understand a visualisation is thus to inquire into its provenance and into the social work that it does. It is to note its principles of exclusion and inclusion, to detect the roles that it makes available, to understand the way in which they are distributed, and to decode the hierarchies and differences that it naturalises. And it is also to analyse the ways in which authorship is constructed or concealed and the sense of audience is realised. (1)

To inquire into the "provenance" and "social work" of the iconic karyotype in contemporary genetic discourse, there seems no better counterpart than the visible public fetus.

Sophisticated feminist analyses of fetal ultrasound point to some important implications of the role of the visual technology in establishing a "fetal presence" (Berlant 1997; Duden 1993, Petchesky 1987). Photographic and ultrasound images of the fetus have become commonplace in the late twentieth century. "In the last twenty years manipulations of these images...have produced new contexts for their meaning, and new meanings have penetrated without entirely saturating the experience of everyone's reproduction in everyday, ordinary life" (Berlant 1997, 86). In this section I will build an analogy between the image of the fetus and the image of the genome, in an attempt to draw on existing scholarship around fetal ultrasound to interrogate how "gene presence", fostered by its karyotype image, is coming to "penetrate without entirely saturating" our experiences of reproduction, health and life.

There are two stories to be told in my comparison of fetal and gene imagery. First, the practices of making both the fetus and the genome seem autonomous and self-evident, have problematic political and economic implications. These include effacement of bodies and relationships, imbuing the "objects" (fetus and genome) with the status of cultural symbols, and sanctioning projects of domination, alienation, surveillance, and normative manipulation in reproductive "management". In a more promising and less simplistic vein, I will argue that, like the visual image of the fetus, the genome image is part of a specific "phenomenon" whose production includes material

constraints, apparatuses, and a particular cultural and historical context (Barad 1998). Reconceptualizing the "fetus" or the "gene" as a dynamic intra-action rather than a thing-in-itself allows possibilities for reworking the phenomenon in ways which include "acts of subversion, resistance, opposition and revolution" (Barad 1998, 33). My analysis will speak through Barad, Haraway, Petchesky and Berlant to examine the ways in which fetal imagery can inform a theoretical approach to meanings being simultaneously stabilized and challenged by gene imagery.

The visual characteristics of the fetal and gene image are strikingly similar. Haraway describes the fetal image as "free-floating, anatomically sharp, perspectivally registered" and "self-evident at first viewing" (1997, 178). Similarly, karyotype images contain morphologically distinct chromosomes in a two-dimensional plane, free-floating and clearly distinguished from their vacant background. Their similarity makes them not only a useful exploratory tool for my argument, but an interesting commentary on how they may refer to one another.[12] If the fetus suggests an illicit look into a mother's womb (Petchesky 1987), the collection of chromosomes may, when associated with prenatal diagnosis, imply a view inside the cell inside the fetus inside the mother. They could be interpreted as continuous images in the life cycle from the single fertilized cell to the developing embryo. Following the conventions of an already established tradition of fetal imagery, the gene image becomes more self-evident to experienced viewers. We see what we have learned to see. Like the erasure of the mother in fetal photography/ ultrasonography, karyotypes erase the cellular source of the chromosomes in order to focus on the heroes of the drama.

One of the main feminist criticisms of fetal ultrasonography is this effacement of the mother (Petchesky 1987; Duden 1993; Berlant 1997).

> Prior to the new technology, the mother's expanded body had functioned both as the representation of the fetus's body and as its armor. The

[12] While they are not explored here, I am cognizant that there are some important differences as well. The fetal image, in its likeness to a human form, is evocative of potential life in a way which is different from, although not unrelated to, the karyotype. Furthermore, there is probably more of a convergence in fetal imaging, whereas multiple images come to mind when one speaks of the "genome" - including the double helix, autoradiographs, fluorescent imaging, etc.

> expansion of the fetus to human and even superhuman scale within the frame of the photograph shattered the aura of maternal protection, making the fetus miraculous in a new way...this transformation of representation and scale pushed the mother into the fuzzy, unfocused part of the picture, throwing her body into a suspension of meaning and value with implications both intimate and national (Berlant 1997, 108-109).

The newly visible fetus granted it an autonomous presence which has resonated in abortion debates and courtrooms, where the fetus has become a contested terrain, and an entity often portrayed in conflict with the mother (Petchesky 1987). In hospitals, fetal patienthood has allowed lucrative expansion of prenatal care and fetal surgery (Petchesky 1987). Analogous to maternal erasure, imaging of genes and chromosomes has facilitated a literal and psychic effacement of the cells and bodies from which the genetic material is extracted. Genome autonomy has surfaced in courtrooms in issues of genetic parenthood, criminal culpability and charges of "wrongful life" wherein a doctor can be sued for failure to provide prenatal screening which may have prevented the life (Rapp 1999, 40). Genome patienthood can be seen in glamorized, but as yet severely limited attempts at "gene therapy". Genome presence has channelled vast resources into projects (like the Human Genome Initiative) which attempt to locate health and disease at the molecular level, to the exclusion of social, economic, and environmental aspects of disease causation.

A further effacement in both fetal and chromosomal imagery are the apparatuses (material, instrumental and discursive) of their production. Both the fetal and gene images epitomize "the distortion inherent in all photographic images: their tendency to slice up reality into tiny bits wrenched out of real time and space" (Petchesky 1987, 62). The fetus owes its existence as a public object to visualizing technologies (Haraway 1997, 174). The image has become so ubiquitous that its authenticity is rarely questioned and it is widely accepted as an accurate representation as a "real" fetus, obscuring the processes of deliberate image construction (Petchesky 1987). In a similar vein, I have argued previously that the acceptance of the karyotype as a

transparent look into our cells is an abstraction because it obscures the negotiated practices of laboratory tinkering and image processing inhered in its construction.

There is an absence of an established critical literature around genetic and chromosomal imagery. Fetal ultrasonography, on the other hand, has generated a great deal of commentary from feminist scholars and activists because, from the point of view of abortion rights, its use is more contentious and more overtly political. My analysis suggests that the increasingly ubiquitous karyotype, in its visual and conceptual similarity to fetal images, may be implicated in a political and corporate agenda which favours genetic essentialism. The image makes visible the genome as a thing-in-itself. Gene presence has the consequences of funnelling resources toward molecular biology to the exclusion of alternative approaches to health and disease, allowing commodification of genes and gene products, sanctioning questionable practices such as prenatal sex-selection, and allowing valuation of genetic characteristics to determine whose lives are worth living.

The karyotype image, like the fetal image before it, has come to saturate contemporary experiences of pregnancy and life. The image is embedded in discourses of risk, the normal and the abnormal, the viable and the abortable. Again, like the fetal image, it is an object of the "intimate public sphere" (Berlant 1997), where political debate about stem cells, genetic health and abortion infuse private conceptions of one's reproductive and lived experience. Barbara Duden, in her book Disembodying Women tracks the historical emergence of the visible fetus, locating Lennart Nillson's 1965 images in *Life* magazine as a turning point wherein the newly visible fetus gained an ontological status different from when it was invisibly embedded in a woman's bodily experience (1993). Her exploration of the fetus' "misplaced concreteness" is motivated by a concern about "how a woman's acceptance of this kind of fetus not only disembodies her perceptions, but forces her into a nine-month clientage in which her "scientifically" defined needs for help and counsel are addressed by professionals" (4). Similarly, the karyotype signifies an appraisal of genetic worth, and channels a body whose chromosomes have been imaged into a medicalized understanding of selfhood.

Conclusion

If this were the end of the story, I could clearly be accused of granting the images the deterministic power that I refuse to grant to the "unknowable" objects to which they refer. Fortunately (and always) the picture is more complex than it at first seems. In problematizing simplistic acceptance of the narrative which threatens to reduce liveliness to photos and maps of genes, we can continue to consult feminist critiques of ultrasound imagery. Two insights are particularly useful in destabilizing the fixation of images.

First, neither the images, nor the reproductive technologies they infer, are met with uniform responses. Issues such as experience, race, class, sexual orientation and access belie any simplistic notions of "Everywoman's Reproductive Consciousness" (Petchesky 1987, 71). Fetal images are diversely met with appreciation, resistance and partial appropriation. Some women experience comfort and participation rather than alienation as a result of ultrasonography (Petchesky 1987). Similarly, a karyotype has clear diagnostic uses which are, or have the potential to be, beneficial. The same characteristics which make the image aesthetically appealing may have positive implications for people who wish to identify with their "genetic blueprint". For a fetus not aborted based on cytogenetic testing, the karyotype may appear beside the ultrasound image in a baby book. As Karen Barad points out, lesbian couples using new reproductive technologies are appropriating genetic "advances" to subvert heteronormative codes of parenthood (1998). These examples of resistance do not undermine the need to look critically at implicit meanings being made in western public consciousness, but they do accomplish the important work of preventing the critique itself from reinscribing the hegemonic and victimizing narrative.

A second destabilizing framework, applied to fetal ultrasound, is developed by Karen Barad in "Getting real: technoscientific practices and the materialization of reality" (1998). Barad borrows from Neils Bohr to argue that objects do not have inherent observer-independent properties. They are instead constituted by the intra-action between the material being observed, and the apparatus of observation, which includes the instruments, the human observer, the community to which he or she belongs, the cultural and political context etc. There can be no inherent cut between the material

and the agencies of observation. Instead, they co-constitute a "phenomenon". The measurements and properties yielded by an experiment belong not to the object, but to the entire phenomenon, whose "unambiguous account requires a description of every circumstance of the experimental arrangement in order for the fact to hold "true" (Barad 1998, 20).

Barad's framework is more helpful, for my purposes, when applied to the case of fetal imaging:

> [U]ltrasonography is not an idealized surveillance technology, a merely physical instrument that provides a view of the fetus as it exists independently of observational apparatuses. Rather, ultrasound technology designates specific material-discursive practices, limiting what is seen and produced in accordance with its own iteratively intra-active technoscientific, medical, economic, political, biological, and cultural etc. development as an ever-changing phenomenon... (25)

As I read Barad's argument, fetal presence and maternal effacement is a product, not of the autonomous characteristics of the fetus or of the image, but of a temporal collusion of particular circumstances comprising the phenomenon - including the material inside the woman's uterus, the woman, the piezoelectric crystal inside the ultrasound transducer, the monitor, the technician interpreting the image, the policies which determine who has access to ultrasound, and for what purposes etc.

How can Barad's "phenomenal" analysis help us to resolve the question of how to read, or what to read into, a karyotype? Barad's attention to the apparatuses of production and relations of power suggest that we can locate the fetal image in a dynamic cultural milieu, undermining its apparent fixity. Similarly, I have demonstrated that the karyotype is a node in a chain of representations, and that a great deal of social, aesthetic and instrumental work is implicated in its stabilization. The character of such an image, however, is that in its tangibility, its "thereness", its simplicity, it effaces this ambiguity. This is its beauty: it becomes a scientific object that we can work with, that can help people make difficult clinical decisions, that can be separated from its

clinical context to generate funding for the Human Genome Project. It suggests that we can distil a body into its essence, for public viewing. Barad's analysis, and mine, is both a caution against this reductionism, and a conviction that whatever apparent stability visual scientific objects achieve, it is ephemeral.

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