Essay

The Molecular Genetics Testing Lab

On the Fringe of Personhood

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Abstract This article proposes to consider the deterministic potential of genetic testing by confronting the genetic "mystique" portrayed in popular culture (and in certain scientific literature), in which DNA is seen as the soul of the cell and genes as master molecules (Lindee and Nelkin 2004), with molecular genetic testing laboratory practices. We will look at the question of what genetic testing does, that is, the practice of genetic testing itself. The particular molecular testing laboratory we will be looking at tests for genetic markers associated with DSD (Disorders of Sex Development). The testing process reveals a previously invisible component of the body through the aid of technology, and a complex picture unravels regarding the role genes play in being considered "un-well".

Keywords 5-alpha reductase; bio-sociality; DSD; genetic testing; laboratory studies.

Introduction

Genetic testing (the search for the presence or absence of genetic material) and genomic testing (the search for factors that may encourage the expression or action of genetic material, Dupré 2004) have entered the field of medicine in numerous ways (O'Malley and Dupré 2005; Lindee 2005; Ankeny and Parker 2002). Genetic and genomic markers can indicate a family history of biologically-linked diseased (as opposed to purely environmentally-linked), can help understand if organ donors should be of similar or mismatched ages (based on mRNA levels), or can indicate the possible variation of developmental pathways in the body, among many other diagnostic practices.

In this article we will be looking at genetic testing in relationship to developmental pathways. More specifically, we will be looking at how this biological data is framed in the context of the laboratory setting and laboratory practice. We are interested in the potential "special status" of DNA/genetics, and the use of deterministic versus systemic models in the framing of genetic material.

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Greatly simplifying, we can describe two extremes of biological models: the deterministic model in which a singular biological component is believed to determine complex biological and/or social factors; and the systemic model in which the body and social factors must be understood as interactional and mutually dependent (regarding genetics see Allen 2002; Portin 1993). The particular laboratory we will be looking at tests for genetic markers associated with DSD (Disorders of Sex Development, referred to as Intersex syndromes from 1917 to 2006; Dreger and Herndon in morland 2009, pp. 205-209). The genetic/biological data found can shift the gender-assignment of a very young individual, and therefore has some very strong implications for the individual's life path and experience of embodied identity.

The relatively new aspect of this biological information in clinical settings (in our case from 2000) begs numerous questions. The testing process reveals a previously invisible component of the body with the aid of technology (Lock, Young and Cambrosio 2000; Clarke *et al.* 2010). Many of the questions that arise revolve around the biological developmental model of the body (deterministic or systemic), and the identity implications of the DSD diagnosis. How is the genetic data framed in the lab? Is it taken to be determining factor in forming the body? How is it interpreted regarding identity factors such as gender identity? Is it seen to be a biological piece in a larger complex puzzle? What is the role of laboratory practices in influencing the significance given to the biological data? What are the implications of the varying positions?

The practices in the lab, aimed at individuating a specific genetic marker that is directly linked to diagnostic nomenclature, points to a deterministic framework. The genetic marker equals the syndrome. And yet, a complex picture unravels regarding the significance of both the syndrome and the role genes play in being "un-well". As a colleague suggested, the genetic testing itself emerges as an artifact that participates in a complex web of techno-scientific practices. Interpretations of the genetic data vary from patient to physician, and from discipline to discipline. We will be looking at the overlap of interpretations in this particular genetics lab, which veers from the deterministic model one might assume.

1. Genetic testing, identity metaphors and laboratory practice

Genetic testing raises a red flag in a multitude of disciplines because it is assumed to propose a biologically deterministic model of personhood and pathology. What we are talking about is the conceptual difference of being genetic diseased (marked as defective in the presence of genetic variance; Billings, Rothstein and Lippman 1992), potentially un-well due to the statistically probability associated with the genetic marker, or simply diverging from the statistical norm regarding a genetic marker (with or without associated biological or social "problems"). Disability theory warns of a new eugenics (Taussig, Rapp and Heath 2003; Shakespeare 2005), in which pre-natal genetic testing could be

used to eliminate undesired and/or socially stigmatized (and often misunderstood) biological difference.

Rose and Novas (2004) discuss the concept of biological citizenship, pointing to patient groups where group identity is based on a biological aspect, such as a genetic marker (variation in a genetic sequence), or a genetically related disease. The genetic marker is pictured as representing a biological entity that has a special status above other biological markers (blood type, hormone levels, etc.), somehow deeply tied to identity. The potential special status of genetic markers has lead policy makers in various nations to propose bioethical guidelines that regulate genetic biobanks as if genetic material were different than other biological material[1].

Genetic material contains information that potentially (symbolically and biologically) refers not only to the individual but also their family (through hereditary markers) (Clayton 2003). This consideration, combined with the special status given to DNA (and genetics) as a primary biological marker in explaining personhood, makes genetic material seem especially sensitive and personal. DNA has rapidly acquired vast symbolic currency in contemporary society, interpreted as the "book of life", or the biological key to who we are (Lindee and Nelkin 2004). The public image of genetic information is often biologically deterministic, relating to individual, family and group identity.

The term biologically deterministic can mean two things: a theory that interprets life from a strictly biological point of view; or a theory that proposes biological factors determine how an organism (such as people) develops, behaves, interacts, etc, to the exclusion of social and/or environmental factors. Popular discourse will often utilize a deterministic image of genetics, transferring the rhetoric of heredity, shared family traits and behavior, to this biological marker: "he's hot headed, it's in his genes" (Lindee and Nelkin 2004).

Biological scientists, however, claim the charge of biological determinism is a simplistic accusation, seeing as a large part of contemporary genetic research looks directly at the interaction between genes and the environment. And yet, biological determinism is the explanatory key between the subtly differing concepts of *being* genetically diseased, seen as unhealthy, flawed, pathological, and *having* a genetically linked syndrome, seen as a possible difference in the development of the organism which may or may not affect the function of said organism.

In this article we will approach the genetic testing (and biological determinism) debate from a different angle; the laboratory practice of genetic testing. In this manner we can directly observe if and how deterministic theories

[1] UNESCO International Declaration on Human Genetic Data (2003), http://www.eu-patient.eu/Documents/Projects/Valueplus/Patients_Rights.pdf, accessed 11/03/2011; Additional Protocol to the Convention on Human Rights and Biomedicine, concerning Genetic Testing for Health Purposes (2008): http://www.jus.uio.no/english/services/library/–treaties/03/3-04/genetic_testing.xml, accessed 11/25/2011.

play out in the practice of genetic testing. Geneticists often propose a systemic biological framework, identifying genetic markers as important biological information that is dependent on an interactional system. For instance, epigenetics looks at the environmental factors, such as heat or timing that affect the manifestation or expression of genetic material, declassifying the genetic material as the determining agent; Evo–Devo genetic theory focuses on evolution and development, yet again proposing a multi-dimensional model regarding the relation of the genotype (genetic composition of the organism) to the phenotype (composite of an organism's observable characteristics or traits) (Jablonka and Lamb 2005). Both of these biological theories dismiss neo-mendelian deterministic models that claim one gene directly represents one biological (or social) trait. If genetic material is declassified from "the book of life" to part of an intricate whole, it loses its potential as the new eugenic threat.

This article proposes to consider the deterministic potential of genetic testing by confronting the genetic "mystique" portrayed in popular culture (and certain scientific literature), in which DNA is seen as the soul of the cell and genes as master molecules (Lindee and Nelkin 2004), with molecular genetic testing laboratory practices. We will look at the question of what genetic testing *does*, that is, the *practice* of genetic testing itself.

As originally proposed by Kuhn (1962), laboratory practices reveal the boundaries of the scientific habitus, and thereby the rationale that creates the practice. Latour and Woolgar (1979) argue that by observing scientific practice we are not discussing whether a scientific fact is valid, but what scientists (and the network of actors involved in reinforcing a scientific fact) think this fact *does* and means. The *meaning* of the scientific object is where the scientific "fact" is transformed into a social object and practice (Latour 1987). In the molecular genetic laboratory, the digital bio-data results of the testing processes are translated into the social realm when practical significance is given to the material being manipulated. Genetic test results in-of-themselves have no innate meaning, they acquire meaning in context.

We hope to demystify the hidden meaning attached to DNA in social discourse in and out of the lab. Medical practice essentially reflects a *useful* model of biological theory, aimed at achieving a specific result. Genetic testing is aimed at finding a biological marker that hopefully inserts itself into a therapeutic protocol that better serves the patient. At the current state of technology genetic testing primarily serves as a diagnostic tool. By achieving a more accurate diagnosis one hopes for better medical care.

Whether the genetic material is interpreted deterministically or systemically can greatly alter the therapy model offered to the patient. In our case, it can also affect the gender assigned to the patient. In addition, how the genetic information is communicated greatly changes the interpretation, or stigmatization, of the diagnostic category. We worry about biological determinism in genetic testing for two primary reasons, the potential threat of a new-eugenics (the elimination of potential humans due to genetic/biological variance), and the conceptual reduction of complex traits such as identity and

behavior to a handful of bio-data. However, are these deterministic concepts part of the theory embedded in molecular genetic laboratory practices? The answer itself is somewhat ambiguous. A deterministic vision can inform the rationale to perform genetic tests, and in turn, inform their interpretation. In some cases the genetic data will shift the deterministic model from some other part of the body to itself. Simply, the genetic artifact is given meaning through a complex web of techno-scientific interactions informed by numerous theories regarding biology, the body and their social relevance.

2. The power of representation

The symbolic power of the gene, DNA and genetic medicine have been explored by Susan Lindee and Dorothy Nelkin (2004), who claim that the "DNA Mystique" has captured the medical and public fancy to a point where the genetic component of a cure or research program *in itself* becomes a marker of validity. This is possible because DNA is portrayed as the symbolic biological *locus* of heredity, the passage of traits from one generation to the next. People often say: "it's in his genes", when someone acts like their parents or family. In molecular biology the passage of complex traits is believed to be an intricate process involving much more than just DNA. However, symbolic logic pushes DNA, and genes, to represent even complex social traits such as behavior and identity.

Lindee and Nelkin argue that genetic symbolism is powerful because it fits so easily into other social metaphors: that kinship is in the blood, that race is biological, that people have "natural" abilities, that physical disability is a sign of overall dysfunction, and so forth. They are quick to point out that these social metaphors are not based on scientific facts, but *use* scientific facts to reinforce the naturalization of social inequality. The overlapping symbolism in eugenic discourse and genetic testing makes the terrain of what genetics *means* and *does* uneasy.

Lindee (2005) discusses the positivist rhetoric surrounding genetics in *Moments of Truth in Genetic Medicine*, rhetoric that offers genetics as a potential miracle for every ailment. Genetic medicine is currently primarily genetic testing, which offers itself as a diagnostic tool that does not add any new therapeutic option to pathology treatment. However, diagnosis itself can be a fundamental aspect of treatment. Lindee points out how patient groups will lobby for genetic research, feeling that they are not being taken seriously otherwise. A genetic marker can put a disease or syndrome on the map of pathologies, creating funding systems, attention, etc. The genetic marker, however, has the primary function of imbuing pathology with an added biological reality. With a genetic marker one can say "I have this" with certainty, as opposed to referring to a set of symptoms.

There is a part of genetic rhetoric (and practice) that is inherently deterministic. The gene was conceptualized, before it was actually considered a

physical entity, as a biological unit of heredity (Morgan 1935). It was proposed as key feature in dictating development. Yet, from the very beginning of what we consider genetic research, the deterministic power of the gene was ambiguous. Genetic research flowered in the fields of agriculture and animal husbandry, where both line-purity and advantageous mutations are sought (Theunissen 2008). Experiments in creating a productive product in these fields (before and after genetic theory) had always highlighted the possible combination of negatively perceived traits with positive ones. In addition, early drosophila fly experiments indicated the role of environmental factors (timing, heat, etc) in gene expression or phenotypic development. The gene was given a dominant and *necessary* role in development (Maienschein 1984), yet there were always other factors to consider.

Of course this symbolic dance with undisputable biological *truth* and identity is what makes the genetic discourse so interesting and tricky. A genetic marker may often aid a linguistic shift from saying, "I *have* this syndrome" to "I *am* this characteristic" as can be the case with mental illnesses and physical differences (I have/am schizophrenic/disabled etc.). Based on the social use and/or prejudice surrounding a medical diagnosis, patient groups might seek or shun genetic testing. In both cases, the genetic marker is imbued with the power of the final truth of biological explanation (Rapp 2000).

Since genetic testing was introduced in DSD diagnosis[2] genetic markers associated with certain syndromes have become biological markers that indisputably *confirm* the presence of said syndromes. The genetic data will generally trump other biological data in the choice of gender assignment. Depending on the position taken by the physician, the genetic data can be seen as more relevant than other aspects of physical gender presentation or expressed gender identity (in older patients).

In some cases the genetic personhood metaphor has been extended to include complex social traits such as behavior and sexual identity. Popular science reporting is rife with discovery of genes for bi-polarism, homosexuality, compulsive behavior, and so-forth. Many molecular biologists argue that it is currently impossible to find a singular biological marker for complex traits, that may or may not have biological components, such as behavior. Utilizing Lindee and Nelkin's argumentation, we could imagine that it is the DNA mystique itself that creates research funding for projects that are potentially scientifically unsound and have no therapeutic value. A prime example is the search for the homosexual gene.

On a lesser scale, DSD patients have seen much funding moved towards identifying genetic markers. Italian DSD patient group members (AISIA and KIO[3], representing respectively Androgen Insensitivity Syndrome and Kline–felter's syndrome) have participated in genetic data collecting for the euro DSD network. While one AISIA member is intrigued by her genetic status (she has a

[2] In the year 2000 for the Italian university hospital used as case study in this article.
[3] http://www.aisia.org/home.html; http://www.klinefelteronlus.it/

pen-pal in Canada with the same genetic marker), she wonders how this genetic information is going to help her with the issues associated with the syndrome, such as adequate hormonal replacement therapy, sterility and the social stigma. She and others in the group hope that a new reading of genetic variance can help reduce the stigma associated with DSD. KIO founder echoes AISIA's concerns, hoping that money will be put towards quality of life research, such as on the health effects of hormones. He and other members of KIO indicate that increased chromosomal testing has helped reduce the stigma of Klinefelter's syndrome specifically because it has shown how common it is (estimated at 1:700 male live births; Fausto-Sterling 2000, p. 53).

Genetic testing can be broken into two primary categories, prenatal and post-natal. Pre-natal testing carries with it the negative association with the eugenics movement and the moralization of normality. Nikolas Rose (2006)[4] discusses the nuance of genetic diagnosis as being "potentially unwell", highlighting the link between the predictive nature of genetics and identity. In a similar manner Margaret Lock (2005) refers to the increase of genetic testing as the new divining, a new diagnostic tool that indicate probabilities, much like the ancient Greek oracles. Pre-natal testing reflects not only our expectations of what technology, or bio-medicalization, should be able to do for us (Ettore 2000), but also the expectation that we *reject* a perceived imperfection (Rapp 2000). Ryna Rapp postulates that this "modern divining" (Lock 2005) incurs social pressure *to do something* about this advanced knowledge. Rapp indicates that potential mothers will be shamed or held accountable for choosing to continue a pregnancy where prenatal testing has revealed a genetic variance associated with syndrome categories.

3. The power of representation. Visualizing molecular genetics.

The laboratory setting we will be looking at instead deals primarily with post-natal testing. Therefore the eugenic threat is an unpleasant shadow that has already been avoided. The genetic markers in question, that we will meet in the next section, evoke Rose's conception of bio-sociality. The genetic markers are laden with the *potential* for the individual to be un-well, as well as implications regarding identity. The genetic markers sought by this specific laboratory have, in a relatively short time, wed themselves with the definitions of the syndromes they represent. The markers therefore affect the identity of the individual, *and* the identity of the diagnostic category.

In genetic testing, DNA is visualized, converted from an invisible component in a blood sample to a visible digital representation. As Luc Pauwels (2005) reminds us, these scientific visualization practices seek not only to render the invisible visible, but also to provide a scientifically useful representation of the biological material. DNA material is converted into bio-data through a complex

[4] Building on his work with Carlos Novas (2000).

series of processes that involve chemical additives, light wave technology and electro-processes. One of the final steps in genetic testing, genetic sequencing, utilizes DNA electrophoresis to separate DNA fragments by size. The end result of this process visualizes the DNA strand as a digital list of letters that represent the nucleotide sequence.

Genetic testing (in its many guises, from adult diagnostic testing, to pre-natal testing, to forensic testing) provokes a wide variety of debate and conflict of opinion, which can be considered on two axes. The scientific axis questions the accuracy and utility of a mechanistic representation of genetic material. The social axis questions the relationship of DNA to personhood and identity. Can a digital representation of biomaterial really tell us who we are, what is right or wrong in our body, whom we came from? The reductionist image of DNA irks our sensibilities surrounding our complex sense of identity, yet it also irks branches of science that insist on a complex model of the organism.

Due to the complexities of development, in certain DSD cases, the "sex" chromosomes (XY,XX) do not "determine" the sex of the individual, let alone their gender. Biological sex has come to be simplistically represented by the sex chromosomes since their "discovery", alternatively represented by the gonads, the genitals, or secondary sex characteristics throughout history. US 1920s and 1930s "sex" hormone research indicates perhaps more accurately that biological sex is the *total* impression of the differences in male and female bodies (Rechtor 1997). The genetic marker linked with a given syndrome is associated with the development of all the biological components of sex, as well as the statistical probability of gender identity.

Genetic testing superficially seems to offer a biological model, which follows the neo-mendelian 'one-gene one trait' model, implying a deterministic and mechanistic vision of DNA, life and the body. This is in contrast with epi-genetics and other branches of molecular biology that view genetic material as part of a systemic process, in which the mere chemical structure of nucleotides does not in itself "code" for anything if taken out of its specific biological context (Jablonka and Lamb 2005). Epi-genetics points to simple factors, such as temperature and timing, which can drastically change the development of an organism while maintaining the same genetic material. Epigenetic, but also bioethical, historical and sociological discussions around the practice of genetic testing question the limits of the mechanistic model of genetics (Ankeny and Parker 2002). The sociological critique mirrors the epi-genetic critique; that life cannot be encapsulated in one biological process (Lippman 1991; Goodman, Heath and Lindee 2003).

In most cases, genetic testing is not seeking to mechanistically define the individual through its genes, it is instead looking for a genetic marker that will confirm what the medical team already thought was the case based on anecdotal information and other symptoms. Finding the genetic marker of a suspected syndrome can greatly aid treatment by canceling-out the use of dangerous or useless therapies. That DNA, genetics, and genomics have taken on more symbolic meaning than the materials themselves can actually provide or perform

is beyond a doubt. The reification of genomic information has lent itself on one hand to a positivistic faith in what this information can provide for humanity, and on the other, a plethora of bioethical quandaries about how to deal with the rise of the new quantities of biological data being gathered and stored.

The scientific visualization process of DNA proposes genetic material as an important biomarker, worth both the economic and temporal investment. Yet it also proposes DNA as an inert object, which must be manipulated in order to be visualized and interpreted, and therefore qualified as bio-data. The DNA manipulation/visualization process is mechanistic, expected to produce consistent repeatable results. Testing for specific genetic markers is also atomistic, in that it practices the belief that biological objects are important and relevant separate from the organism and separate from their dependent biological processes (Allen 2002). And yet, that the biological entity is expected to be consistent and atomistic in a mechanistic testing process, does not imply that the mechanism of the biological entity itself is expected to be atomistic and deterministic.

4. Creating Data

This description of the average process of molecular genetic testing comes from a two-year period of intermittent observation in a University Hospital in Italy. I alternately shadowed the four team-members through their daily routine, as a participant (note-taking, question asking) observer. I charted the arrival of several patient cases/blood samples from their arrival to the communication of the test results/diagnosis to the team physician. I also charted the testing phases, the interaction between the lab members, and the interaction with the larger DSD team. Through situational analysis (Clarke 2005), I hoped to decipher what the team members thought they were doing. What they thought was the aim of the testing procedure, what was a good result, good practice, but also what they thought the role of this bio-data was in the overall treatment procedure. Beside this particular focus on the molecular genetics lab, I also frequented Italian DSD patient groups, and conducted in-depth interviews with other members of the DSD team.

The lab I frequented is a primary Italian lab that tests for a handful of genetic markers that indicate certain DSD (Divergence/Disorders of Sex Development) syndromes. The lab can be considered primarily indicative of the testing protocol for these genetic markers, secondarily of Italian laboratory practice. As Mol (2002) indicates in her own research, this laboratory setting is neither exemplary nor unique to the national context, but provides interesting insight into the practices involved.

This lab receives blood samples from all over Italy, rendered doubly anonymous through a coding system. Molecular testing became routine for DSD in this university hospital in 2000. Since then, the DSD team has been expanding their research on the other DSD health factors implicated by the genetic markers.

At this point, however, molecular testing primarily supports diagnosis accuracy and corresponding gender assignment. They test for 6 genes that are implicated in CAH (Congenital Adrenal Hyperplasia)[5], AIS (Androgen Insensitivity Syndrome)[6] and 5-alpha reductase (Syndrome name and genetic marker are the same)[7]. As we will briefly discuss later, the molecular testing has had the unexpected repercussion of diminishing irreversible non-consensual childhood surgery (one of the bioethical hotspots in DSD treatment), specifically in 5-alpha reductase and PAIS[8] (Partial Androgen Insensitivity Syndrome) diagnoses. from the 1950's onward most centers throughout the world adopted John Money's Optimal Gender of Rearing (OGR) care model (Dreger 1999; Fausto-Sterling 2000; Karkazis 2008). In Money's model social factors such as the childhood rearing environment trump biological factors in the establishment of gender identity (a model which was greatly appreciated in the 70's as it seemed to favor social determinism). However, Money saw the genital *form* as being the most important factor in influencing the rearing environment (unambiguous treatment as one gender or another) and established the protocol of early childhood genital surgery (preferably before the age of three to avoid memory of the experience) (Dreger 1999; Karkasiz 2008) that also lead to a policy of secrecy in which the patient (and at times the parents) was left in the dark regarding their diagnosis and treatment. Unfortunately genital surgical techniques often require

[5] CAH indicates the hyper-activity of the adrenal gland, leading to a high production of steroid hormones (such as hydro-testosterone), that can lead to: salt wasting in some forms; in XX children mild to severe masculinization of the genitals in-uterus, or after birth; early on-set puberty; unusual hair growth. This syndrome is clinically subject to the highly controversial early childhood genital surgery (to de-masculinize the genitals, similar to clitorectomy) and stigmatization of "ambiguous" genitals. It has also suffered clinically from the confusion and erroneous overlap of concepts such as gendered behavior, gender stereotypes (especially regarding energy levels and aggressive play), gender identity, and sexual identity. Varies from 0.35% of Yupik Eskimos to 0.0005% New Zealanders with an estimated average of 0.00779% (Fausto-Sterling 2000). As with most DSD syndromes CAH was subject to a legacy of secrecy, lack of informed consent and shame.

[6] AIS indicates the insensibility to androgens in a XY individual. In the complete form the individual will have "male" gonads and "female" genitals and secondary sex characteristics (1:13,000). In the partial form the genitals may be considered "ambiguous" and subject to early childhood surgery (1:130,000). This syndrome is subject to gonadectomy for psycho-social (not functional) motives, vaginal lengthening surgeries (now dilation is offered) and stigmatization due to the belief that XY chromosomes "means" a person is a man. AISIA is the Italian patient group. http://www.aisia.org/home.html.

[7] 5 alpha-reductase, is caused by a deficiency in the enzyme 4 steriod 5-alpha reductase. In the Dominican Republic it is known as Guevedoche (lit. balls at twelve), due to increased and different forms of androgens at puberty that cause the body to "masculinize". In cultures where this syndrome is common, some individuals raised as girls retained a female gender identity, however most take on a male gender identity (more advantageous in the social hierarchy, Herdt 1996, p. 437). In western bio-medical culture this syndrome is thought to lead to the development of a male gender identity (see Hertz 1996).

[8] Often used as a catchall diagnosis, once very diffused, now primarily in the absence of a genetic marker.

maintenance or repair (e.g. dialation of the vaginal canal), implicating numerous medical visits and examinations often in front of numerous medical students (critically described as medical stripping; Morland 2009) (Dreger 1999), of course children/patients intuit that something is "wrong" with them, and/or their genitals (leading to shame and stigma; Morland in morland 2009, pp. 285-312). In addition, most patients were assigned the female gender, simply because the female genitals were considered "simpler", as a noted surgeon stated it was "easier to dig a hole than built a pole" (Hendricks 1993, pp. 10-16; Dreger 1999).

There is little space in this context to discuss the ethical conundrums of DSD treatment[9], while the entrance of molecular testing into care protocol has had interesting and unexpected repercussions. The gender assignment implications underlying DSD diagnosis highlight the identity aspects of the genetic discourse. Medical curiosity surrounding gender in the body has often had reductionist/deterministic overtones, focusing on one component of the gendered body (such as the gonads or genitals) or another. In contemporary biological models of sex there is debate and controversy over the developmental pathways of biological sex, and the *locus* of sex (that is: the factors that are considered to be most important in swaying the gendered body to develop in one way or another).

At the end of the nineteenth century, hundreds of theories of sexual differentiation could be documented, but by the 1920's all theories would take into account sex chromosomes and sex hormones (Maienschein 1984, p. 457). DSD syndromes displace sex chromosomes as the primary organizer of sex in the body, and since their very conception, researchers looked deeper for the mechanisms leading to sex determination. Already in 1927 the Danish geneticist Øyvin Winge proposed that there must be a 'testis determining factor' on the Y-chromosome, which was linked to the development of the male phenotype (Holme 2007, p. 152).

Genetic markers, in conjunction with hormones and hormone receptors came to be seen as responsible for disrupting the one to one relationship between chromosomal sex and phenotypical sex (the fully developed type or the external appearance of the body). The phenotype is then believed to represent the gender identity of the individual. It is still often popularly believed that the sex chromosomes make one "really" a man or a woman. Shifting the "real" indicator of biological sex from the chromosomes to genetic markers does not entirely depart from a deterministic rationale, yet leaves some space open for a systemic, interactional model.

The genetic marker, in the case of a suspected DSD, is subject to a diversity of explanatory models that range from reductionist to systemic. It is important to keep in mind that the genetic test is performed *when a diagnosis has already been proposed*, and the genetic marker serves to confirm or adjust the suspected

[9] Primarily regarding nonconsensual childhood genital surgery, lack of informed consent, medical stripping, stereotyped idea about both social and physical gender, etc. See Dreger 1999; Fausto-Sterling 2000; Karkazis 2008; http://www.isna.org/index.php

diagnosis. If a genetic marker is found, the diagnosis acquires a higher level of indisputability. If it is not found, other anecdotal and biological information will support the diagnosis. While the genetic marker trumps all other biological material in diagnosis assessment, it is not necessarily taken to determine the development pathway on its own.

As is the case in most medical genetic laboratories, in this lab the technician already knows what they are looking for before they start the testing process. They are specifically asked by the medical team or collaborating hospital to look for the genetic markers associated with the suspected syndrome, therefore they are not directly involved in the diagnostic decision process. The anecdotal and physical data acquired in medical interviews with the patient have already led the medical team (in this hospital led by a pediatric endocrinologist) to suspect a diagnosis, or a potential genetic marker. For instance, several AISIA members have been re-diagnosed from PAIS to different syndromes such as 5-alpha reductase or Leydig Cell Hypoplasia[10]. there has been a general effort to use genetic testing to clear-up earlier ambiguous diagnoses.

One AISIA member had a difficult process digesting her renewed diagnosis as 5-alpha reductase, having long accepted (or at least digested) her PAIS diagnosis and the subsequent negative surgical experience (resulting in almost total lose of genital sensation). She has a female gender identity, non-stereotyped gender behavior and a homosexual orientation. She mourned the possibility that she could have been raised a boy and avoided the type of medical treatment she received, however, after a year or two, she decided she was happier as a woman (despite and because of her experiences)[11]. In the past, non-stereotypical gender behavior and/or homosexual orientation would cause the medical team to reevaluate the gender assignment. The very different categories of gender identity, gendered behavior, gender appearance and sexual orientation are still often confused or overlapped. Historian Elizabeth Reis indicates throughout the medical obsession with then termed pseudo-hermaphroditism in the 17th and 18th hundreds doctors would often put aside the gonadal information (then considered to be the biological determining factor) in order to affirm a gender assignment that rendered the individual heterosexual (doctors had the authority to influence the assignment of legal gender status).

5. Creating Data. Diagnosis

[10] Leydig Cell Hypoplasia is a condition resulting from reduced or absent functioning of Leydig cells which leads to insufficient production of androgens, which can affect sex differentiation.

[11] While there are no conclusive statistics, it appears that there is a higher instance of transgenderism (transition from one social gender category to another) in the general population than among those diagnosed with a DSD, especially since the protocol of assigning most patients the female gender has been revised.

The lab team searches for the genetic marker that has been indicated by the physician. The combination of the identified related genetic pattern results and the tacit knowledge of the technicians leads to either a positive or negative result, there is no grey-scale interpretation of data that may or may not reflect a scientific paradigm[12]. However, each team member may have their own interpretation of what the test results *mean*, regarding diagnosis and the gender identity of the patient.

The laboratory procedure tries to isolate the molecular component that is associated with the diagnosis they are leaning towards. I accompanied different technicians through the steps that lead to the isolation of the genetic marker, who were clearly experts in laboratory procedure, not necessarily in gender or social theory. I was shown how to extract, purify, determine the concentration of, and then amplify the DNA. It certainly seems like a miracle to render DNA sequences visible, through this cleaning and replication process. It also requires a lot of patience. Throughout the various processes we added chemicals and centrifuged, taking always-smaller samples, rendering what had once looked like blood into a clear liquid like water. The DNA is then read and analysed for the specific marker that is being looked for. Hidden in the blood is the significant biological object that will be read. However, this object must be manipulated in several ways and even boned with other chemicals before it is palpable as useful data.

One blood sample will go through the same procedure several times, to test for the different suspected markers but also to guarantee the accuracy of the result. One blood draw provides enough biological material to perform multiple tests, and leave stored material for future use. Blood arrives from all over Italy, or by foot from an adjacent building. The day I arrived, in fact, we received blood from a local source that had already been coded to protect the patient's identity. The only remaining identifying factor was the suspect diagnosis.

One of the technicians brought me to the ward where they took the blood samples, four beds in a room, and on the way, we passed the psychologist and head endocrinologist, with the family of a child with a 5-alpha reductase diagnosis. This family had a hard time coming to terms with multitude of explanatory models they were offered by the medical team and the society at large. They originally wanted to maintain the female gender assignment (due to genital size) and modify their child's genitals to seem less "ambiguous", following Money's OGR model. However the medical team suspected and then confirmed the 5-alpha reductase diagnosis, which made them push for a male gender re-assignment. The 4-year-old child in this case was included in the process to some extent, and knew that their gender was considered "unclear", and would ask which bathroom they should use. The psychologist later indicated that the child did not clearly indicate a gender preference, yet their stress symptoms (jaw clenching) greatly reduced when the finally male assignment

[12] See Turrini (2011) for a discussion of variable visual representation *versus* digital representation.

decision was made (remaining unclear if the stress alleviation was due to the end of ambiguity and intense medical attention, or the male gender assignment).

The 5-alpha reductase diagnosis, through the visualization techniques of the molecular genetics lab, changed the child's life in many ways: from the medicalization techniques he will live through, to the gender he was assigned by the medical team. Equally importantly, this diagnosis led the medical team to advise against irreversible genital surgery and attempt less invasive methods. The child started topical genital androgen treatments to increase the size of the genitals, thereby immediately avoiding sensation reducing surgical techniques, hopefully leaving him the decision to have, or not have, genital surgery at a later date[13].

6. Creating Data. Laboratory Practice

Back in the lab, to extract the DNA we took 3ml of blood and added a patented solution (Cell Lysis Solution) to break the cells. I found it very interesting how much of the testing process was standardized outside the lab, through patented formulas and machinery with specific protocols. These patented processes, of course, still leave room for individual tacit knowledge in practice. Each technician had their area of specialty, their tacit knowledge and their quirks. My first informant had been with the lab for 30 years, from before the time in which you needed a specialized degree to be a molecular lab technician, and he was a local. He explained to me the progression of DSD chemical diagnosis techniques, and abandonment of others, from radioactive processes to siphoning chemicals like one does with gasoline. They used to search for sex hormones and growth hormones, now they look for genetic markers.

My first informant made it very clear that he thought the most important thing in the lab was to be good technician, which is to be clean, organized and thorough. He was not particularly interested in the latest genetic theories. He seemed to portray the idea that the lab techniques were all similar in the end; machines, solutions and protocol changed, but the process was the same. Joking, he answered my questions as to why he did certain things with a little rhyme, "non so per che cosa, so fare le cose" ("I don't know why we do things, I know how to do things")[14]. This was obviously ironic, because he had little things to say about everyone, and every technique. He had been in the lab longer than many others, mastering the techniques as they changed. He implied that he always handled the extraction due to his precision, the others (who all had

[13] Many patient groups advocate the delay of all irreversible early childhood genital surgery, indicating this intimate procedure, with its many side-effects, must be decided by the individual/patient. This decision is supported by the Italian National Bioethical Committee (Comitato Nazionale di Bioetica 2010) but is not part of Italian medical protocol or law.

[14] This direct quote is awkward in Italian.

specialized degrees in genetics) left things a mess, an obstacle to accuracy. There were glass jars everywhere, like a glassmakers workshop, but everything was sterile with surgical plastic inside. Disposable products place the responsibility of sterility on the manufacturer, removing it from the lab.

It was like returning to college chemistry: titration (drip), and centrifugation. Every step used different droppers with differing levels of accuracy, and different centrifuges for differing sample sizes. The first (extraction) process broke the cells to extract the DNA, through the use of a chemical solution and the centrifuge. The second step purified the DNA with a second chemical solution (Nuclei Lysis Solution) and again the centrifuge. One needs to know how to unpack DNA by inviting the unwanted material to separate away. Besides the glass jars, we had entered into the world of standardization and patents. The choice of the *right tools for the job* (Clarke and Fujimura 1992), that is the scientific justification of instruments and protocols, are increasingly being decided outside of the laboratory, by manufactures and increasingly international protocols. Each machine came with a brochure, pre-mixed chemical solutions and a protocol. This repetition of standardization evokes the mechanistic nature of the laboratory process.

For instance, we purified with a Wizard® genomic DNA purification kit. As we followed the instructions from the kit, however, I found every step had its own non-written tacit-knowledge aspect: agitate like this, it should look like this when it comes off the bottom, etc. This tacit knowledge displayed an intimate relationship to the *visual* aspects of DNA in its various manipulated forms, each of which are different forms of readable data. The first several rounds of centrifugation left the blood sample red, a clot floating in the CLS, which is dispersed and then put back together through the aid of a protein solution. Another round of the centrifuge cleans away the red blood cells and we were left with a clear liquid.

The first "miracle"[15] of DNA visualization is performed by Isopropyl alcohol (C3H8O) that reconsolidates the material, and you can see the DNA floating on the bottom of the plastic vile. That is, you have created something you can look at under a microscope. To the layperson it would just look like a little dirt in water. For the technician it is already bio-data, potentially useful information. When you remove the liquid there is a little substance that seems like tiny strands of cotton. The cleaning process is replicated with alcohol and then the DNA is re-hydrated. The samples are then kept in different fridges based on their properties.

On a different day in a different room we determined and amplified the DNA. The previously cleaned sample is "read" by a 260/280 nm wavelength. When DNA is isolated from organisms, frequently some protein remains present in the DNA solution. Protein is tightly bound to the DNA and the complete removal of protein is not always possible. To determine the concentration and purity of the DNA solution, the absorbance of UV light is measured in a spectrophotometer.

[15] As described by the first technician.

Both protein and DNA absorb UV light, but they have different absorbance curves. The peak of light absorption is at 260 nm for DNA and at 280 nm for protein. When you run a spectrum of absorbance with varying wavelength, you should see that both curves slightly overlap in the area between, and including, 260 and 280 nm. Thus, when a solution contains both protein and DNA, absorbance at 260 nm is mainly due to the DNA present, and a little bit by the protein. At 280 it is the other way round. By dividing the two absorbance-values, one can calculate the purity of the DNA solution. These barely visible cotton strands of DNA are visualized in yet a different way, as light absorption, yet this bio-data has no practical application, it needs to be further manipulated.

In the amplification process different enzyme primers are added to a standardized chemical mixture in a process called the Polymer Chain Reaction, which multiplies the chain to seem infinite[16]. The polymer chain reaction method relies on thermal cycling, consisting of cycles of repeated heating and cooling of the reaction for DNA melting, and enzymatic replication of the DNA. 70° C opens the molecule, at 95°C the primer attaches itself, and at 68°C the chain forms. Primers (short DNA fragments) containing sequences complementary to the target region, along with a DNA polymerase (after which the method is named), are key components that enable selective and repeated amplification. As PCR progresses, the generated DNA is itself used as a template for replication, setting in motion a chain reaction in which the DNA template is exponentially amplified. PCR can be modified to perform a wide array of genetic manipulations[17].

The technician indicated the importance of writing everything down and checking each step, so as to not forget anything. The protocols they applied, beyond the protocols in the brochures, seemed aimed at regulating human fallibility, techniques that made sure you incorporated every step, with little room for variability. These first two technicians (one trained in genetic theory and one not) seemed to have little interest in the meaning or the result of their practice (limiting themselves to comments about the importance of an accurate diagnosis), yet they were very proud of their technique, their craftsmanship. The important role the genetic-data has in the diagnostic process is on some levels taken for granted.

The steps in the visualization process indicate an intricate understanding of the materiality of genetic data, how it will behave in certain environments, how to isolate it, how it is made. It was hard to identify any specific genetic theory in the visualization process, whereas many other scientific theories were at play, like thermodynamics, basic chemistry etc. The prepared solutions are complemented by a control and a water sample. Technicians often use their own bio-mater in the control process, as a way to make sure they have not contaminated the samples.

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[16] As described by the second technician.

[17] Description synthesized from written lab instructions and oral instruction.

The amplified DNA is purified by yet another patented process, using the QIA quick spin kit and the QIAquick Nucleotide Removal Kit. The slogan in their instruction pamphlet reads "making improvements in life possible!". The patented kits included these small bursts of propaganda in their instruction manuals, which reflect the "DNA mystique", however the technicians used them to explain to me why there were not separate lab guidelines. The kit even includes the right size tubes for the machine so there is no need to have separate lab supplies. The technician counted as he laid out the samples in the machine with the buffers, indicating that everyone develops different methods to make sure that they have not skipped any. The tacit knowledge employed in every step seemed directly related to maintaining accuracy and purity of the samples, that is basic lab techniques, as reflected by the observations of the first lab technician. At this point we had 20 samples for every patient tested. The plastic vials had gotten so small there is nothing left visible or even imaginable to the naked eye.

At this point the extracted, purified, determined, amplified, re-purified DNA is loaded on the agarose gel and "data voltaggio" (literally: given voltage). This is where the physical entity of the DNA falls away and is transformed into digital data. The electrogram exploits what we know about charges in molecules to move and order them for measurement and visualization. As in all of the previous processes, chemical or electrical manipulation of the DNA is a means to an end, an essential part of the process, yet not essentially part of the bio-data itself. These manipulations of DNA have the aim of rendering DNA visible, palpable and useful. The assumption is that the essential material of DNA, what it needs to communicate to us, is not changed in any way by these processes, but rather, exposed and emphasized. The genetic data does not seem to acquire special status in the lab practice, yet requires many special instruments adapted to fit the specific purpose.

The final result of these chemical electrical manipulations is the series of letters we have come to associate with nucleotide sequences, or genetic patterns. Two technicians spend the rest of the afternoon reading the sequences to each other, first to identify possible contamination or mistakes, then to compare the sequences to "normal" sequences, and already established variant sequences that are associated with certain syndromes. The technicians who read the electropherogram are not just well trained technicians capable of recognizing errors in a long string of letters, they are also well trained in genetic theory.

It is only in this last step that the technicians begin to express opinions about the relevance of the genetic bio-data. In fact, these last two technicians have more direct interaction with the DSD team, and potentially the patients. They are the first to tell you that a genetic marker indicates a spectrum of development possibility, not necessarily a problematic pathology. The meaning they give to the test results is primarily empirical: the digital data says these are the genetic markers present in this part of the DNA. Underlying this meaning is the belief that this digital data will help the medical team treat the patient by giving a more accurate diagnosis.

However, contextually to their hospital team, they give another meaning to the bio-data. Critical of past paternalistic protocols that hid diagnosis and treatment options (including non-surgical options) from the patients (and often their parents too), they read the bio-data as an empirical entity that empowers the patient. They see the genetic test as inherently linked to new protocols of informed consent and full-disclosure, no longer something to be ashamed of and hide. The bio-data is situated as symbolically more modern, technologically advanced, and thereby associated with more modern standards of patient care. The bio-data they provide is linked to a body of scientific literature (easily found on the internet by the patient or family) that avoids stigmatizing terms like pseudo-hermaphrodite, assumes full disclosure to the patient, and contextualizes genetic variance.

The experimental process, and the creation of biodata, is definitely mechanistic and reductionist. It certainly could seem to reflect a biologically deterministic model. And yet, the genetic maker simply indicates a diagnosis, and therefore a pathology (a statistical deviance from the norm), but not necessarily a disease (a disturbance in the organism that incurs dysfunction and/or suffering) or a problem (Billings, Rothstein and Lippman 1992). The difference lies in the interpretation of the genetic material.

7. From data to meaning

The communication of genetic test results relays meaning onto the digital rendering of the DNA. As we saw in the beginning of the article, the scientific debate regarding genetic testing reflects the interpretation of genetic material as either independent/mechanistic or system-dependent. The social debates further question the role of biological variation in disease and identity definition. In the last ten years the new figure of the genetic councilor has been instituted to explain genetic data to the patient. The genetic councilor often translates seemingly determinist digital genetic bio-data into the language of genetic probability and possibility.

This particular DSD team does not have a referring genetic councilor. The genetic test results are communicated by a physician, generally a pediatric endocrinologist. The lab's head geneticist told me that many parents (and adult patients) end up calling her directly to ask for further information and explanation of the genetic data, yet she does not have an official role in diagnosis communication. The geneticist implied that the other doctors (not trained in genetic testing) are more likely to portray the genetic results as deterministic (neo-mendelian, one gene=one trait) biological truths, leading the patient to believe certain *dysfunctional* symptoms will *definitely* manifest. There is a distinct difference between reading genetic variance as linked to physical difference, and interpreting that difference as inherently dysfunctional.

This geneticist's personal opinion is confirmed by research on termination rates in Klinefelter's syndrome diagnosis. Klinefelter's syndrome is a DSD

syndrome that is silently targeted in prenatal cytogenetic testing, evidenced by a third sex chromosome (XXY). Termination rates were found to be much higher, in three different geographic and cultural settings, when the diagnosis was communicated by a gynocologist, pedatrician or general practitioner, than when the communication was conducted by a genetic councilor (Abramsky *et al.* 2001; Hall *et al.* 2001; Hamamy and Dahoun 2003; Yon-Ju *et al.* 2002). These authors explain their findings by proposing that a genetic counselor is more likely to explain genetic indicators as representing a varied spectrum of development than non-specialists, as well as having more updated information about genetically-linked syndromes. As genetic testing has found its way into increasing disciplines, an increased percentage of "invisible" (not particularly symptomatic) Klinefelter cases have been revealed. Genetic councilors accuse non-specialist practitioners of promoting not only a deterministic model, but also a model that over-pathologizes genetic variance. The Italian Klinefelter's patient group (KIO) promotes genetic research *because* they feel it will show how common and diverse the syndrome is.

There can be an understanding gap between popular conceptions of neo-mendelian genetics, and molecular genetics that relies to some extent on the developmental model. The geneticist must explain two factors that have emerged in molecular genetics, the complex model of development that goes beyond the chromosomes, and the difference between a genetically-based syndrome and being un-well. Molecular genetics represents the genomic paradigm, in which the performance of the genes and their interaction with non-genetic factors are the objects of research. The genomic concept has difficulty mapping directly onto the dualistic social model of gender. This philosophical issue regarding the de-moralization of biological variance[18] can be instrumental in helping patients understand and accept a previously unheard of difference.

The practical work of the genetics lab plays out in various ways: diagnosis communication (in this lab), statistical evidence of development and molecular markers, implications for postponing early irreversible interventions. Molecular testing is generally performed after birth, thereby the bioethical debates such as fear of eugenic elimination practices can be limited to chromosomal prenatal diagnosis and not molecular genetic testing as of yet. The geneticist of the lab said, "Parents call me asking, 'they've found this genetic marker, what does it really mean?'". Genetic counselors are appearing in certain medical fields (such as the cancer ward of this hospital) but ironically not always in this sensitive arena where adults/parents must make decisions for children/patients.

The other implication of molecular testing for this lab is gender assignment, the focus of so much of DSD medicalization. Molecular testing provides much greater accuracy in diagnosis, even though even the geneticist indicated that many people diagnosed with DSD do not have any of the established genetic

[18] See Feder in Morland 2009, pp. 225-247 for a historical/philosophical description of the evolution of the morality of physical variance from the eighteenth century in regards to DSD, but also Foucault 1979, pp. 177-184.

markers. However when the genetic marker is present, it will distinguish the diagnosis from the once catchall category of PAIS (Partial Androgen Insensitivity Syndrome). Historian and biologist Ingrid Holme (2007, p. 2) wonders:

> Yet as the historical analysis of the shift between the one sex to two sex model indicates (Laqueur 1990), it remains to be seen whether the social sphere will respond by incorporating this new evidence into the tacit, everyday understandings of sex or seek to maintain the binary and fixed relationship(s) between men and women by governing them as males and females.

In a previously mentioned case, molecular testing revealed a 5-alpha reductase genetic marker, changing the original PAIS diagnosis. This case, among others, gave weight to the members of the DSD team who opposes irreversible early childhood genital surgery. In this case the parents' dis-ease (Kleinman 1986) with their child's non-standard body was medicalized through counseling and hormones instead of irreversible surgical manipulation. The belief in Western biomedicine that 5-alpha reductase indicates a male gender identity directly shifted care protocol in two key manners: the proposed acceptance of a boy child with a micro-phallus, and the advice to postpone surgical intervention until the patient is self-determining. The *locus* of gender identity was to some extent defined by the molecular genetic marker.

Vernon Rosario (2009) hypothesizes that the complexity of genetic expression promoted by molecular research will lead to an equally complex model of sex and gender that he calls quantum sex. However, historian Garland Allen (2002) references his own difficulty in relaying a non-mechanistic or non-deterministic model of genetics in teaching upper-division college students. The one gene=one trait model is inaccurate, but easier to understand. The professional use of genetic counselors may help in the diffusion of a non-deterministic model.

In fact, even experts sometimes express opinions that reflect the influence of appearance, behavior and phenotype on what they think about a patient's genetic make-up. I heard contradictory comments in some cases, for instance, in the case of an XY adult, one technician commented, "poor thing she thinks she's a lesbian, but really she's a man". The patient had an uncontested female identity throughout her life, combined with female sexual object choice. This same technician firmly believes that XY individuals with Androgen Insensitivity Syndrome are women. Yet, the patient in question had a mixed molecular marker similar to 5-alpha redutase that is associated with potential male gender identity in the western bio-medical context. This technician will insist that XY chromosomes do not make you a man, yet sometimes a molecular marker is taken to indicate the same authority that chromosomes once did in gender determination.

Despite occasional opinions that could be perceived as deterministic, the geneticists generally advocate for a complex, developmental model. This genomic model generally refutes the deterministic language of the 'gene for x social trait', but rather, as Fox-Keller (2000) suggests, views genes as processes. The lab

technicians, in fact, seem to interpret their digital data as part of a complex process, while outside of the lab this data is somehow flattened to represent something in-and-of-itself. New genomic research continues to affirm an increasingly inter-relational model of sex development. As Holme (2007, p. 171) indicates:

> The view of the body as an active process is widespread in the discussions of the paradigm shift from studying single genes in genetics to studying genetic networks in genomics (Moss 2003).

In the hospital laboratory individual genes are targeted for very practical reasons in order to promote more accurate diagnosis.

8. Conclusion

> Visual representations in science differ significantly in terms of how they relate to what they purport to represent (i.e. their representational and ontological status). Visual representations in science may refer to objects that are believed to have some kind of material or physical existence, but equally may refer to a purely mental, conceptual, abstract constructs and/or immaterial *entities.* (Pauwels 2005)

The visualization of hidden biological components is part and parcel of DSD diagnosis. Technology has helped shift the *locus* of biological sex to parts of the body that would otherwise remain unknown, invisible. The visualization processes that convert blood samples to electropherograms and genetic digital data are standardized procedures that invoke a myriad of scientific theories and techniques, as well as the social metaphors that DNA represents. By taking a walk through the actual practice of genetic testing we can see that the commitment to the deterministic model implied by the practice is ambiguous. The laboratory practice relies on the assumed predictability of chemical interactions, aided by heat, speed, light and electricity.

By digitally visualizing DNA we are manipulating its material support, as well as its potential and its meaning. The DNA mystique, the positivistic rhetoric surrounding DNA and its cultural symbolic value has induced the *need* to visualize DNA in ever-increasing settings (Lippman 1992). In this manner, it seems the increased practice of genetic testing relies to some extent on deterministic assumptions such as the special status of DNA and genetics in describing the body. However, geneticists indicate that they see this information as only one part of the puzzle.

The practice of genetic testing treats genetic material as a physical chemical entity, which can be manipulated in many ways, without losing its informational value. In fact, it must be chemically and thermically manipulated in order to reveal itself. This would superficially imply that genes are believed to be resistance to external influences, however, the laboratory manipulations hopes to

"clean away" interfering biological information. Genes are initially read atomistically, separate from the organismic context, then inserted into a systemic explanatory model. The explanatory model of the molecular laboratory proposes that genes are partially deterministic, in that genes determine part of development in interaction with other biological processes. However, the primary purpose of the genetic lab is to *confirm* a diagnosis, not explain developmental processes. The genetic information has the explanatory power to support or negate a suspected diagnosis, such as Klinefelter's syndrome or 5-alpha reductase, but does not indicate *how* the syndrome will manifest. The increased diagnosis of these genetically linked syndromes lends statistical evidence to the *variety* of manifestations of these syndromes.

I would argue that the molecular genetic labs practices reflect the belief that genes have an important biological *potential*. That is, in certain biological conditions, the genetic marker will lead the body to develop in a divergent direction, and therefore it is important in the medical context to identify suspect markers to anticipate what *might* happen in the body. There is no strong deterministic paradigm in the lab that indicates that the genetic marker creates individual identity or an un-well individual. The lab tends to adopt the potentially un-well model, indicating that a patient *has* a genetic marker or a syndrome as opposed to *being* genetically diseased. This would indicate that the strong deterministic interpretation of genetic material is created in social discourse and other scientific discourse, not in the lab.

Historians and philosophers such as Lindee (2005) and Moss (2003) highlight the divergence of the scientific practice and social discourse. They indicate the myriad of things that genetics and genetic medicine *cannot* do or describe yet, from creating cures to biologically describing behavioral traits. Lindee in particular indicates that the actual science is far behind the positivistic rhetoric surrounding genetics, while indicating that patients themselves sometimes create these expectations.

As long as the "genetic mystique" reigns in the public image, accompanied by the neo-mendelian deterministic model, genetic testing can be a potential eugenic threat, as well as a tool to stigmatize biological difference as "not right". However, this interpretation is influenced by how genetic information is described to patients, and how patients interpret this information. The deterministic platform is not entirely reflected in laboratory practice. As genetic testing becomes routine in an ever increasing number of medical fields, time will show us if the strong deterministic model continues to dominate the public image of DNA and genetics, or if perhaps genes will slowly lose their special status, becoming a biological marker among many.

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