

Paternalism does not work!

Education Does.

by Sophia Siedlberg
December 2000

Did I ever mention the deep confusion that can be felt by a teenage girl who walks into her local G.P.'s surgery with a small but significant worry about not menstruating? There she is, in front of her G.P. and, as calm and as matter of fact as you can get, he comes out with "You have an XY chromosomal pairing, you are a man, not a woman" and the next thing you know the girl is taking a walk to Beachy Head (seaside cliff on England's south coast where people commit suicide). Oh what an admirable bedside manner! This story is becoming less common. It has been known to happen though.

As for the Science

A great many misdiagnoses were made on this basis? For example we have the buccal smear test, it is quite simple. You take a smear from the lining of the mouth, you look for a Barr body (the result of an inactive X chromosome) and if it is there it is logical to assume the individual is "genetically female". So simple. Well no, it is far from that simple. Back to our woman in the G.P.'s surgery, she has some form of A.I.S. most likely "Grade 6 or 7 A.I.S." (as she presents as a female with little or no confusion before now). It is the simplistic and arbitrary labels and tests that have been so endemic within the medical profession for so many years that are to blame, and most importantly the way they have been used. In theory, you can have a woman, with most internal female organs, who has the 46XY pairing.

The "Y" chromosome is only a part of the story when it comes to resulting in a male phenotype. The SRY gene, (about 4k-bases in length) is seen as the only gene responsible for producing a male phenotype, reducing something as complex as sex differentiation and gender identity to one gene. Anyone who studies genetics will tell you that attributing complex elements of the human condition to a single gene is simply bad science. I can think off the top of my head of at least 10 genes for which there are alleles that can effect the expression of SRY to such an extent that would render SRY useless. Then again, if a gene called KIAA0396 did not express, the differentiation from an initial female phenotype would not take place. Were this gene and SRY compromised, you could theoretically have a female, with a uterus, and partial ovaries but also with "XY" chromosomes. In plain English the potentially female bits would not die off in a developing foetus and the male bits would not develop. O.K. that is a bit theoretical and really the preserve of specialists in that field.

The theoretical possibilities are limitless, in practice they do appear. P.A.I.S. is a condition that affects me, I was born an "intersex" and that essentially is that. Yet this seems to cause society a lot of discomfort. "Such births cannot happen." Very helpful if you are a parent of such an individual or are such an individual yourself. There is a "grading" system for people with A.I.S. as this is really an umbrella term for numerous conditions that "feminize" an "XY male" (to use the paternalistic line). But we really need to look at it another way. In essence it is quite simple, there is Grade 1 A.I.S. (fully male with a few open ended hormone issues) to grade 7, (just next to the KIAA0396 extreme). So if a KIAA0396 and SRY mutation were found, that would be Grade 8, I imagine (though grade 8 does not appear to exist right now). To me this is a bit of a "Pheno meter" (from phenotype = body form, as opposed to genotype = genetic makeup) when put in the hands of the wrong people - such as paternalistic medical professionals. With the wrong approach it could become like the "Phallo meter" (the dubious practice of "pre-sexing" an intersex child with a tape measure placed against the enlarged clitoris/undeveloped penis).

There is a serious point here though, it isn't the grading system that upsets me, it's the degree of "perceived maleness or femaleness" it may engender, how some people may misuse it. For me at least there is a streak of scientific purism in my outlook. Take my external phenotype for example. It spans

across the scale. You can look at one attribute and put me in one place on the scale, another attribute puts me somewhere else. I am going to be open and honest about myself. I never had acne, (Grade 7) my pubic hair distribution was a little sparse, no hair under my armpits, and pelvic hair is certainly not all that prevalent (Grade 7). Ah but I had a little facial hair! (Grade 1 - spiralling crash into hell). I am tall, slim, well endowed in the breast department but with small nipples (I fly back to grade seventh heaven) and mixed genitalia, (down a few floors). I have seen numerous leaflets with scales like this. And without a visit to an endocrinologist who knows exactly how to apply this system, or a patient group who have adopted it. I hover up and down on this label roller coaster feeling very, very insecure. The perhaps disconcerting debate here is how far you are from the "expected status" of someone with an XY pairing.

The real issue for me is perhaps the 46XY thing. It is like some blot in my universe. Haunting me with that acrocentric, SRY wielding self hate inducing monstrosity "that chromosome". Or is it? In fact when you look at it from a genetic standpoint you may be surprised. I want to concentrate on a variant of P.A.I.S. because it is perhaps the condition that is most fraught with negative assumptions. Believe me, as a patient I find myself constantly at odds with people. It is perhaps best to explain in more depth exactly what it is that makes the differences on a genetic level. I will discuss my own condition as a reference. "5 alpha reductase deficiency". This is not the same as "full blown" A.I.S. (meaning that the causal variables were different that is all). But it is a classic example of what mathematicians call a "butterfly effect". Essentially the gene that produces 5 alpha reductase, needed to convert Testosterone to Dihydrotestosterone (which is more potent) needs to be "correct". If the gene (this is an interesting case for the single gene merchants) is compromised, the result is either intersex or outwardly female. With "5 alpha reductase deficient A.I.S." Missense mutations occur when a single base in a codon (3 base triplet) is swapped for another. So the codon does not code for the correct amino acid in a polypeptide chain.

OK., in English:

There is, as I said, a gene. This is a portion of DNA composed of four "bases" - these are themselves "amino acids" denoted A (adenine) T (Thymine) C (Cytosine) and G (Guanine). or (ATCG). When they make "polypeptide chains" three letters or triplets (such as TTT) will code for an amino acid in the polypeptide chain. A daisy chain of amino acids if you like, of which there are 20 to choose from this time. These are the main subunits of proteins and enzymes. TTT as DNA codes for the amino acid "Lysine", for example. A missense mutation is where one letter in the triplet is replaced by another. So for example if "TTT" becomes "TCT", as DNA it will code for another amino acid called "Arginine". So what *was*

H-Serine-Alanine-Alanine-Lysine-Glutamine-NH₂ (DNA = AGACGACGATTTGTT)

becomes

H-Serine-Alanine-Alanine-Arginine-Glutamine-NH₂ (DNA = AGACGACGATCTGTT)

A section of DNA that is active is known as an "exon". So with one variant of 5 alpha reductase deficiency what would otherwise be categorised as a "male" can in fact be categorised as an intersex or female. A transition at the second nucleotide of codon 85 in exon 1 (GGC --> GAC - quoted as DNA not m RNA) substituting glycine for aspartic acid, can occur. (Vilchis F, Mendez JP, Canto P, Lieberman E, Chavez B. 2000). Now this is just one amino acid's difference from the "norm", one base on the nucleotide and one resulting amino acid in the enzyme (polypeptide chains make an enzyme) rendering it non expressive. There are tens of thousands of bases in some genes, the alteration of one results in such a drastic variation. It certainly puts the Y chromosome on shaky ground as a reference point for "maleness". More interesting is the fact that the gene that is involved (SRD5A2) occurs on chromosome (pair) 2, an autosome, not a sex linked chromosome.

Now there are many variations of 5 alpha A.I.S. and this is down to two things. How the expression of other alleles (variation of a given gene) are effected and what the allele of SRD5A2 is. There are many

alleles of SRSY1A2, some with missense mutations in different places along the gene. The interesting point to make with this example is that the sex is effectively decided by an autosome, not a sex chromosome.

Other types of AIS involve mutations in many different genes, some in the X chromosome, others (like mine) in the autosomes. The positive thing for any A.I.S. individual who feels comfortable as a woman is that no one can say that she is “genetically male”. In fact no one can say, “The sex chromosomes make you male or female by themselves”. They are subject to the final say of genes lying in the rest of the chromosomes. The interesting point though, when discussing the “single gene” argument is this. We are looking at a single “letter” in one gene radically altering the “expected outcome” but it has to be seen in context. The manifestation of this depends on the activities of other genes. You cannot say, “This gene will always have this outcome”. All you can say is, “This gene expresses to this degree” and if the conditions are right for such a tiny effect to be made manifest then this gene is at the root of the “butterfly effect”. Paradoxically this in itself undermines the idea of single gene dogmas, and thus undermines the notion that SRY, for example, will always result in a male phenotype. All because the effects of other genes need to be considered. A “polygenic context” if you like.

The Gender Agenda

How many times have I heard someone saying that A.I.S. women must really think like men because they have 46 XY. I remember hearing one woman with A.I.S. saying, “Oh I hate it when some doctors find out that I am XY. They expect me to want to jump up and play football or something”. Well she had pointed out the greatest urban myth about women with A.I.S. that is cited. SRY does not code for gender identity. It codes to the differentiation of gonads into testes. Another interesting (contradictory) quote for those who believe that gender identity is biological and “hardwired”. A few years ago something from an unlikely source took gender identity out of the XY domain altogether. Now I know that a number of intersex people find comparisons between themselves and the transgender community can be a cause of distress or unease (it often winds me up when someone tries to define me as transgendered) but something I found interesting happened a few years ago. Something about the way evidence seems to take this elusive “gender identity” away for chromosomes 45 and 46. Many in the transgendered community were going on about the size of a part of the brain called the “bed nuclei of the Stria Terminalis”. This was because the study by professor D.F. Swaab in 1995, on a number of transsexuals brains during post mortem, produced evidence of the size of the BSTc being concurrent with gender identity. The smaller it was the more female the gender identity. Much of that “brain sex” stuff came from these studies as well.

Having said that, the studies conducted by Professor Swaab in 2000 have produced something that is perhaps relevant to A.I.S. women confronted with negative assumptions about their “gender” identity on the basis of the XY myth. Enabling them to reply to the “XY = male mind” myth. A lot of Professor Swaab’s critics challenged him to explain why the size of the BSTc was different between males, females and transsexuals. They suggested that the medication taken by transsexuals caused a change in the size of the BSTc. Well hormones are involved, but during foetal development. In 2000 when all the transgender politics moved on, Professor Swaab came up with a possible answer to that question. By this time transgender was no longer the issue. But the stuff about A.I.S. women being “man minded” sadly still persists.

Well the latest information to come from Professor Swaab gives the following hypothesis. That the anterior portion of the BSTc undergoes apoptosis (a number of cells within that region die off). Androgens actually seem to inhibit this process of cell death giving a larger (let's say masculine) BSTc. The absence of certain androgens and lack of receptors can cause the apoptosis to continue. So what if you are insensitive to androgens to the degree where this mechanism is affected? Personally I think that gender identity is more complex than that. But the “XY = male mind” myth simply does not stand up to the evidence. It is a paternalistic myth.

I may as well be honest, I don’t like football but I don’t attribute that to some tiny part of my brain or my specific genetic make up. I don’t like football because I find it boring! something that both the transgender

community or the paternalistic practitioners of medicine don't seem to understand. But the idea of gender identity when applied to intersex people cannot really be attributed to either nature or nurture. If anything, that is the core of my argument about the gender agenda. People don't seem to listen to anyone without having tried to find an explanation that divorces an individual from themselves in some way.

If I have a point to make here it is not that I don't entertain Professor Swab's research, quite the opposite, I find it interesting. It is that I find the way people read it soon upsets me, as it often translates into some kind of medical policy. Even with the best intentions any study can have very unfortunate consequences. For example, Dr John Money and Professor Milton Diamond. The argument with these two is simple. Dr John Money decided one day to have a theory, and published a paper which says that "Gender identity is social" and then proceeds to assign every infant male and intersex patient with even the most minor of problem to female by surgical means. It is naturally an outrage. There is no consent, just a form of genital surgery being arbitrarily undertaken. Some years later these children grow up and a number of them are not happy about what was done to them. But then what happens? Enter Professor Milton Diamond. It is classic. "These unfortunate boys were forcibly feminised. They are boys!" (Prof. Diamond says, quoting Dr Reiner, who ascribes to the "XY = male mind" idea). Now there is evidence beginning to emerge, slowly, that a larger number of intersex children were wrongly assigned to male than were thought, as well as the fact that many are unhappy with that. But the implication now is that because it is difficult to masculinize an infant's genitalia (true) no such cases will exist, (but they do) and surely they would be happy to have been boys because these victims of Dr John Money identify as male. (So a number of intersex people find themselves dumped into the "transgender ghetto").

In all fairness to Professor Milton Diamond, he is now, I believe, looking into those who were wrongly masculinized. His basic premise in his paper "Pediatric Ethics and the Surgical Assignment of Sex" is that unless there are pressing clinical reasons infant genital surgery is best avoided. Despite the misleading media claims of him thinking only of the wrongly feminised, his paper includes all forms of infant genital surgery.

A Question of Context

The difficulty arises, not from the basic science but how it is understood and used. Back to the G.P.'s surgery, where the G.P. has the woman back in as she expressed how upset she was at his insensitive remarks. She sits there waiting for a clear explanation of what he meant by that remark. He could quote all the good labelling methods, the bad labelling methods. He could insist on theories that are proven or unproven. He can say whatever he likes, however good or bad the science is behind it. But how she feels about herself has been deeply compromised, that is very obvious. Paternalistic attitudes, especially when quoting scientific terms cannot and will not work.

My G.P. is thankfully the opposite. She is a female G.P. but I don't think that is really the issue here. She lets me read some of the documentation and tells me what I need to know. Admitted, having had training myself I probably have a number of suggestions and a more than average level of input. But having said this many patients do go to great lengths to find out exactly what they have. They look up all the literature available. The internet, while far from perfect, does give more information. Paternalism states that the patient is a passive entity. Their body, if you like, is property of the medical profession while the patient is unwell, according to paternalism. Well as Dr John Money and Professor Milton Diamond seem to illustrate only too well. While Professor Milton Diamond is trying to give control to the patient, it does seem sometimes (due to his being mis-quoted) to be on terms dictated by unknown variables that have been translated into biological determinism, or "XY" determinism. Dr John Money had laid down the damaging myth about upbringing determining an individual's "gender identity". If only people could understand that ownership of oneself and the principle of consent are paramount, and most importantly let no one tell you that you are something that is alien to you. What you know of yourself is what the underlying reality will be. It may seem a bit of a cliché but how you live is what matters, not someone else's idea of what your chromosomes are supposed to be saying, or how your upbringing was conducted. That applies to everyone. in all sections of society.

Thankfully there are now some centres of clinical excellence, e.g. a unit at the University College of London that does a lot of research and ethical patient care. But there is more than that. Patient groups are becoming more aware. Even individuals. I myself write bio-informatics software, and often my condition becomes part of my work. So hopefully in the not too distant future, the ignorant paternalist in the local practice will have all the information to hand, information that is also put into proper context.