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How common is intersex? A response to Anne Fausto-Sterling. *Leonard Sax.*

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Anne Fausto-Sterling's suggestion that the prevalence of intersex might be as high as 1.7% has attracted wide attention in both the scholarly press and the popular media. Many reviewers are not aware that this figure includes conditions which most clinicians do not recognize as intersex, such as Klinefelter syndrome, Turner syndrome, and late-onset adrenal hyperplasia. If the term intersex is to retain any meaning, the term should be restricted to those conditions in which chromosomal sex is inconsistent with phenotypic sex, or in which the phenotype is not classifiable as either male or female. Applying this more precise definition, the true prevalence of intersex is seen to be about 0.018%, almost 100 times lower than Fausto-Sterling's estimate of 1.7%.

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Sometimes a child is born with genitalia which cannot be classified as female or male. A genetically female child (i.e., with XX chromosomes) may be born with external genitalia which appear to be those of a normal male. Or, a genetically male child (XY chromosomes) may be born with female-appearing external genitalia. In very rare cases, a child may be born with both female and male genitalia. Because these conditions are in some sense "in-between" the two sexes, they are collectively referred to as intersex.

How common is intersex? In her 1993 essay, biologist Anne Fausto-Sterling acknowledged that "it is extremely difficult to estimate the frequency of intersexuality" (Fausto-Sterling, 1993, p. 21). In this paper we will focus on establishing how often intersexual conditions occur, and what conditions should be considered intersexual.

In her most recent book, *Sexing the Body: Gender Politics and the Construction of Sexuality* (Fausto-Sterling, 2000), Fausto-Sterling maintains that human sexuality is best understood not as a dichotomy but as a continuum. She bases this assertion on her beliefs regarding intersex conditions. A chapter subtitled "The Sexual Continuum" begins with the case of Levi Suydam, an intersexual living in the 1840s who menstruated regularly but who also had a penis and testicles. Fausto-Sterling writes:

While male and female stand on the extreme ends of a biological continuum, there are many bodies, bodies such as Suydam's, that evidently mix together anatomical components conventionally attributed to both males and females. The implications of my argument for a sexual continuum are profound. If nature really offers us more than two sexes, then it follows that our current notions of masculinity and femininity are cultural conceits.

... Modern surgical techniques help maintain the two-sex system. Today children who are born "either/or-neither/both"--a fairly common phenomenon--usually disappear from view because doctors "correct" them right away with surgery. (Fausto-Sterling, 2000, p. 31)

Fausto-Sterling asserts that 1.7% of human births are intersex. This figure was widely quoted in the aftermath of the book's publication. "Instead of viewing intersexuality as a genetic hiccup," wrote Courtney Weaver for the *Washington Post*, "[Fausto-Sterling] points out that its frequency mandates a fresher look. In one study, intersexuality typically constitute 1.7% of a community" (Weaver, 2000). The *New England Journal of Medicine* applauded Fausto-Sterling's "careful and insightful book.... She [Fausto-Sterling] points out that intersexual newborns are not rare (they may account for 1.7% of births), so a review of our attitudes about these children is overdue ..." (Breedlove, 2000). "Most people believe that there are only two sex categories," went the review in *American Scientist*. "Yet 17 out of every 1,000 people fail to meet our assumption that everyone is either male or female. This is the approximate incidence of intersexuals: individuals with XY chromosomes and female anatomy, XX chromosomes and male anatomy, or anatomy that is half male and half female." (Moore, 2000, p. 545)

This reviewer assumed that Fausto-Sterling was using the term intersex in the usual way, the same way in which Fausto-Sterling herself used the term in her 1993 essay, "The Five Sexes" (Fausto-Sterling, 1993): to refer either to individuals who have XY chromosomes with predominantly female anatomy, XX chromosomes with predominantly male anatomy, or ambiguous or mixed genitalia. This assumption is reasonable, because all the case histories presented in her book *Sexing the Body* describe individuals who meet these criteria (Fausto-Sterling, 2000). However, as we shall see, the 1.7% statistic is based on a much broader definition of intersex.

Fausto-Sterling herself has encouraged the belief that a significant fraction of the population is neither male nor female, but intersex. In an interview with *The New York Times*, she said that "I did some research and we found that maybe 1 to 2 percent of all births do not fall strictly within the tight definition of all-male or all-female.... there is greater human variation than supposed ... [We should] lighten up about what it means to be male or female. We should definitely lighten up on those who fall in between because there are a lot of them" (Dreifus, 2001).

IS HUMAN SEXUALITY A DICHOTOMY OR A CONTINUUM?

Fausto-Sterling's argument that human sexuality is a continuum, not a dichotomy, rests in large measure on her claim that intersex

births are a fairly common phenomenon. Specifically, Fausto-Sterling computes the incidence of intersexual births to be 1.7 per 100 live births, or 1.7%. To arrive at that figure, she defines as intersex any "individual who deviates from the Platonic ideal of physical dimorphism at the chromosomal, genital, gonadal, or hormonal levels" (Blackless et al., 2000, p. 161).

This definition is too broad. Fausto-Sterling and her associates acknowledge that some of the individuals thus categorized as intersex "are undiagnosed because they present no symptoms" (Blackless et al., 2000, p. 152). A definition of intersex which encompasses individuals who are phenotypically indistinguishable from normal is likely to confuse both clinicians and patients.

John Wiener, a urologist, has suggested defining intersex simply as "a discordance between phenotypic sex and chromosomal sex" (Wiener, 1999). While this definition would cover most true intersex patients, there are some rare conditions which are clearly intersex which are not captured by this definition. For example, some people are mosaics: Different cells in their body have different chromosomes. A 46,XY/46,XX mosaic is an individual in whom some cells have the male chromosomal complement (XY) and some cells have the female chromosomal complement (XX). If such an individual has both a penis and a vagina, then there is no mismatch between phenotypic sex and genotypic sex: Both the phenotype and the genotype are intersexual. Yet according to Wiener's definition, such an individual would not be intersex. A more comprehensive, yet still clinically useful definition of intersex would include those conditions in which (a) the phenotype is not classifiable as either male or female, or (b) chromosomal sex is inconsistent with phenotypic sex.

This definition is of course more clinically focussed than the definition employed by Fausto-Sterling. Using her definition of intersex as "any deviation from the Platonic ideal" (Blackless et al., 2000, p. 161), she lists all the following conditions as intersex, and she provides the following estimates of incidence for each condition (number of births per 100 live births): (a) late-onset congenital adrenal hyperplasia (LOCAH), 1.5/100; (b) Klinefelter (XXY), 0.0922/100; (c) other non-XX, non-XY, excluding Turner and Klinefelter, 0.0639/100; (d) Turner syndrome (XO), 0.0369/100; (e) vaginal agenesis, 0.0169/100; (f) classic congenital adrenal hyperplasia, 0.00779/10; (g) complete androgen insensitivity, 0.0076/100; (h) true hermaphrodites, 0.0012/100; (i) idiopathic, 0.0009/100; and (j) partial androgen insensitivity, 0.00076/100. The chief problem with this list is that the five most common conditions listed are not intersex conditions. If we examine these five conditions in more detail, we will see that there is no meaningful clinical sense in which these conditions can be considered intersex. "Deviation from the Platonic ideal" is, as we will see, not a clinically useful criterion for defining a medical condition such as intersex.

The second problem with this list is the neglect of the five most common of these conditions in Fausto-Sterling's book *Sexing the Body* (Fausto-Sterling, 2000). In her book, Fausto-Sterling draws her case histories exclusively from the ranks of individuals who are unambiguously intersex. However, using Fausto-Sterling's own figures, such individuals account for less than 0.02% of the general population. None of her case histories are drawn from the five most common conditions in her table, even though these five conditions constitute roughly 99% of the population she defines as intersex. Without these five conditions, intersex becomes a rare occurrence, occurring in fewer than 2 out of every 10,000 live births.

CLASSIC INTERSEX CONDITIONS

Among classic intersex conditions, the most common are congenital adrenal hyperplasia (CAH) and complete androgen insensitivity syndrome. According to Fausto-Sterling's figures, these two conditions occur with roughly the same frequency: about 0.008/100, or 8 births out of every 100,000. There is no dispute that these conditions are indeed intersex conditions. We discuss them here because some understanding of these conditions is essential in order to perceive how these conditions differ from the other syndromes which Fausto-Sterling includes in the category of intersex.

Complete Androgen Insensitivity Syndrome

These individuals are genetically male (XY), but owing to a defect in the androgen receptor, their cells do not respond to testosterone or other androgens (Boehmer et al., 2001). As a result, these individuals do not form male genitalia. Genetically male (XY) babies with this condition typically are born with a vaginal opening and clitoris indistinguishable from those seen in normal female (XX) babies. In almost all cases, the diagnosis is not suspected until puberty, when these "girls" are brought to medical attention because they have never menstruated. Investigation at that point will invariably reveal that these "girls" are in fact genetically male, that they have undescended testicles, and that neither the uterus nor the ovaries are present. These individuals are genotypically male, but phenotypically female.

Congenital Adrenal Hyperplasia

In this syndrome, a defect in an enzyme involved in the synthesis of adrenal hormones leads to a blockage in one synthetic pathway, giving rise to excessive production of androgenic hormones in a different pathway (White, 2001). These androgens will masculinize a female (XX) fetus in utero. At birth, the girl's genitalia may appear completely masculine, or, more commonly, the genitalia will be ambiguous--neither completely male nor completely female but somewhere in between.

ARE THESE OTHER CONDITIONS INTERSEX?

Late-Onset Congenital Adrenal Hyperplasia

In late-onset congenital adrenal hyperplasia, the defect in the enzymatic pathway typically does not manifest itself until late childhood, adolescence, or later, and the degree of disruption is much less than in classic congenital adrenal hypertrophy. Reviewing the list of conditions which Fausto-Sterling considers to be intersex, we find that this one condition--late-onset congenital adrenal hyperplasia (LOCAH)--accounts for 88% of all those patients whom Fausto-Sterling classifies as intersex ($1.5/1.7 = 88\%$).

From a clinician's perspective, however, LOCAH is not an intersex condition. The genitalia of these babies are normal at birth, and consonant with their chromosomes: XY males have normal male genitalia, and XX females have normal female genitalia. The average woman with this condition does not present until about 24 years of age (Speiser et al., 2000). Men with LOCAH present later, if ever: Many go through life undetected or are discovered only incidentally (Holler et al., 1985). For example, if a daughter is discovered to have classic congenital adrenal hyperplasia, the parents often will be tested for evidence of overproduction of adrenal androgens, and one parent thereby may be discovered to have LOCAH. The most common presenting symptom of LOCAH in men is thinning of scalp hair, but even this symptom is seen in only 50% of men with LOCAH under 50 years of age (Dumic et al., 1985).

Fausto-Sterling recognizes that if her definition of the intersexual as "an individual who deviates from the Platonic ideal of physical dirhorphism" (Blackless et al., 2000, p. 161) is to have any clinical relevance, then at least some patients with LOCAH must occasionally have problems which are intersexual in nature. Accordingly, she asserts that "when late-onset CAH occurs in childhood or adolescence and causes significant clitoral growth, it is quite possible that surgical intervention will ensue." (Blackless et al., 2000, p. 161) The only reference given in support of this statement is a first-person account in the woman's magazine *Mademoiselle* (Moreno & Goodwin, 1998). However, the article in *Mademoiselle* describes a phenotypically female but genotypically male (46,XY) individual with androgen insensitivity: in other words, a case of true intersexuality. LOCAH is never mentioned.

In a large-scale investigation of the natural history of LOCAH in women, the chief complaints of symptomatic women were one or more of the following: oligomenorrhea, hirsutism, infertility, or acne. These investigators noted that "in some cases, affected girls have shown mild clitoromegaly, but not true genital ambiguity" (Speiser et al., 2000, p. 527). Many women have no symptoms at all: "Probably many affected individuals are asymptomatic," notes another recent review (White, 2001, p. 25). A recent study of 220 women with LOCAH found mild clitoromegaly in only 10%; moderate or severe clitoromegaly was not reported (Moran et al., 2000).

Sex Chromosome Aneuploidies

Fausto-Sterling defines all sex chromosome complements other than XX or XY as intersex. Specifically, Fausto-Sterling includes Klinefelter syndrome, Turner syndrome, and all other non-XX, non-XY chromosomal variations in the intersex category.

Klinefelter syndrome. Babies born with Klinefelter syndrome (47,XXY) have normal male genitalia. Male secondary sexual characteristics develop normally in puberty, although the testicles typically are small. Erection and ejaculation are not impaired. Most men with Klinefelter syndrome are infertile, but an unknown proportion are fertile (Warburg, 1963). Because Klinefelter syndrome is most often discovered in the course of infertility evaluation, fertile men with Klinefelter syndrome are likely to go completely undetected. Abramsky and Chapple (1997) have suggested that many men with Klinefelter syndrome are never diagnosed because they are phenotypically indistinguishable from normal (46,XY) men.

Turner syndrome. Among the most salient features of Turner syndrome (45,X) are infertility and short stature: Women with Turner syndrome who are not treated with growth hormone typically will be about 16 centimeters shorter than their predicted adult height based on parental heights (Holl, Kunze, Etzrodt, Teller, & Heinze, 1994). Sas et al. (1999) have demonstrated that girls with Turner syndrome can achieve normal adult heights if daily doses of growth hormone are administered. Although most women with Turner syndrome cannot conceive a child, they can carry a child to term if a donated embryo or oocyte is implanted (Hovatta, Foudila, & Soderstrom-Anttila, 2000). Girls with Turner syndrome do not have ambiguous external genitalia (e.g., no clitoromegaly), nor do they typically experience confusion regarding their sexual identity. "A consistent feature documented in Turner's syndrome is the unambiguous identification with the female sex," according to a recent review in *The Lancet* (Ranke & Saenger, 2001, p. 310).

Other chromosomal variants (non-XX and non-XY, excluding Turner's and Klinefelter's). This category includes a variety of sex chromosome complements, such as XXX, XYY, and other less frequent arrangements. Fausto-Sterling considers all such conditions to be intersex. Men with an extra Y chromosome (47,XYY) are not distinguishable from normal (46,XY) men, although the average intelligence of men with this aneuploidy is lower than normal. Their fertility usually is not impaired. They are most commonly discovered in the course of evaluation for mild mental retardation or behavior problems (Fryns, Kleczkowska, Kubien, & Van den Berghe, 1995). Likewise, women with an extra X chromosome ("triple X," 47,XXX) are fertile, although the mean intelligence of women with this aneuploidy is also probably below average (Bender, Linden, & Harmon, 2001). None of these chromosomal variants are associated with ambiguous genitalia, or with any confusion regarding sexual identity. There is therefore no clinical sense in which these individuals are intersex.

Vaginal Agenesis

Fausto-Sterling estimates that about 0.0169 births per 100 are characterized by vaginal agenesis (also known as vaginal atresia), a condition in which the distal third of the vagina fails to develop and is replaced by about 2 cm of fibrous tissue (Simpson, 1999). According to the definition which I have proposed, vaginal agenesis is not an intersex condition. Girls born with this condition have an XX genotype and normal ovaries. In the majority of cases, vaginoplasty restores normal female vaginal anatomy (Robson & Oliver,

2000). Women who have undergone vaginoplasty can and do go on to have successful term pregnancies (Moura, Navarro, & Nogueira, 2000). Nosologically, vaginal agenesis is to genital anatomy as cleft palate is to maxillofacial anatomy. Surgical correction for vaginal agenesis is conceptually no different from surgical correction for cleft palate.

HOW COMMON IS INTERSEX?

Subtracting these five categories--LOCAH, vaginal agenesis, Turner's syndrome, Klinefelter's syndrome, and other non-XX and non-XY aneuploidies--the incidence of intersex drops to 0.018%, almost 100 times lower than the estimate provided by Fausto-Sterling. This figure of 0.018% suggests that there are currently about 50,000 true intersexuals living in the United States. These individuals are of course entitled to the same expert care and consideration that all patients deserve. Nothing is gained, however, by pretending that there are 5,000,000 such individuals.

IS INTERSEX n NORMAL VARIANT OR n PATHOLOGICAL CONDITION?

The most original feature of Fausto-Sterling's book is her reluctance to classify true intersex conditions as pathological. Regarding babies born with both a penis and a vagina, she writes: "Perhaps we will come to view such children as especially blessed or lucky. It is not so farfetched to think that some can become the most desirable of all possible mates, able to pleasure their partners in a variety of ways" (Fausto-Sterling, 2000, p. 113). Fausto-Sterling (2000) strongly affirms her belief that all possible combinations of sexual anatomy must be considered normal:

Complete maleness and complete femaleness represent the extreme ends of a spectrum of possible body types. That these extreme ends are the most frequent has lent credence to the idea that they are not only natural (that is, produced by nature) but normal (that is, they represent both a statistical and a social ideal). Knowledge of biological variation, however, allows us to conceptualize the less frequent middle spaces as natural, although statistically unusual. (p. 76)

NOSOLOGICAL CONFUSION

Nosology is the science of the classification of diseases. The first principle of nosology is the distinction between the normal and the pathological. This principle poses real difficulties for Fausto-Sterling. She often uses the word natural synonymously with normal (for an example, see the previous paragraph). However, natural and normal are not synonyms. A cow may give birth to a two-headed or Siamese calf by natural processes, natural being understood as per Fausto-Sterling's definition as "produced by nature." Nevertheless, that two-headed calf unarguably manifests an abnormal condition.

Fausto-Sterling's insistence that all combinations of sexual anatomy be regarded as normal is reminiscent of Szasz's view of mental illness (Szasz, 1974). Szasz insisted that mental illness is not a real biological phenomenon but merely an invention of society. Like Fausto-Sterling, Szasz was suspicious of the distinction between normal and pathological. Fausto-Sterling follows the example set by Szasz in her belief that classifications of normal and abnormal sexual anatomy are mere social conventions, prejudices which can and should be set aside by an enlightened intelligentsia.

This type of extreme social constructionism is confusing and is not helpful to clinicians, to their patients, or to their patients' families. Diluting the term intersex to include "any deviation from the Platonic ideal of sexual dimorphism" (Blackless et al., 2000, p. 152), as Fausto-Sterling suggests, deprives the term of any clinically useful meaning.

CONCLUSIONS

The available data support the conclusion that human sexuality is a dichotomy, not a continuum. More than 99.98% of humans are either male or female. If the term intersex is to retain any clinical meaning, the use of this term should be restricted to those conditions in which chromosomal sex is inconsistent with phenotypic sex, or in which the phenotype is not classifiable as either male or female.

The birth of an intersex child, far from being "a fairly common phenomenon," is actually a rare event, occurring in fewer than 2 out of every 10,000 births.

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