Intersexes in Humans: An Introductory Exploration

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"SO GOD CREATED MAN in his own image . . . male and female created he them." Gen. 1:27.

> A weird happening has occurred in the case of a lansquenet [soldier] named Daniel Burghammer. . . When the same was on the point of going to bed one night he complained to his wife, to whom he had been married by the Church seven years ago, that he had great pains in his belly and felt something stirring therein. An hour thereafter he gave birth to a child, a girl. . . He then confessed on the spot that he was half man and half woman. . . He also stated that . . . he only slept once with a Spaniard, and he became pregnant therefrom. This however, he kept a secret unto himself and also from his wife, with whom he had for seven years lived in wedlock, but he had never been able to get her with child. . . The aforesaid soldier is able to suckle the child with his right breast only and not at all on the left side, where he is a man. He has also the natural organs of a man for passing water. . . All this has been set down and described by notaries. It is considered in Italy to be a great miracle and is to be recorded in the chronicles. The couple, however, are to be divorced by the clergy.—From Piadena in Italy, the 26th day of May, 1601.[1]

The history of human intersexes[2] extends far back into antiquity. Their existence is probably as old as the species, yet they are not well understood. Few if any societies have been comfortable with the issues they raise. Persons whose sexual identity have been unclear traditionally have been ostracized individually and ignored collectively.

Modern research, turned toward the serious study of intersexuality and related conditions in only the past three decades or so, has found the subject

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to be poignantly complex. This essay is only a brief introduction; it is not possible to explore adequately even one of many specific conditions. Readers interested in more detailed information should consult the works cited in the bibliography.

The very existence of human intersexes poses some interesting unanswered questions in LDS traditions and beliefs. Traditional LDS expressions on gender identity also fall short of embracing the complexity demonstrable in the real biological world. Sex, as traditionally posited, is an immutable characteristic of an eternal spirit, of which the mortal body is only a tabernacle. The body is in the image of the spirit, and it is tacitly accepted that this extends to sexual characteristics. An increasing body of medical data, however, gives one considerable cause to reflect on the precise nature of that relationship. Whatever theoretical role may be ascribed to the influence of the spirit, it is a biological reality that sex determination, in the physical body at least, is affected by and almost certainly controlled and determined by genetic and hormonal means, in other words, it not only has a physical basis of identifiable dimensions, but it is subject to considerable malfunction and reversal.

DETERMINATION OF SEX

In many organisms, both animals and plants, the genetic systems responsible for sexual differentiation (into 0, 1, 2 or up to 10 separate sexes) is well understood, and scores of different systems exist. Sexual differentiation may be controlled by a single gene or gene pair, by complete chromosomes or by purely environmental differences. The system typical of mammals (and thus humans) is basically one of chromosomally-determined sex. The female mammal possesses two special chromosomes designated as X chromosomes. The male typically possesses only one X, but also carries a Y chromosome. In mammalian eggs or sperm (collectively termed gametes), only half the chromosomes are usually present. Eggs produced by the female typically carry one (and only one) X chromosome; sperm may be either X-bearing or Y-bearing. Normally the sex of the offspring is determined, therefore, by the sperm. If fertilization is by an X-bearing sperm, then the resulting XX embryo will usually become a female; if by a Y-bearing sperm, the resulting XY embryo will ordinarily become a male. The system is simple—but fraught with potential malfunctions.

One source of difficulty is that the chromosomes do not always sort themselves out properly during gamete formation. Either excess or insufficient numbers of chromosomes can be packaged into any given egg or sperm. For example, one encounters persons with one X chromosome only—designated "XO." Individuals with other combinations are also known, including XXX, XXXX, XXXXX, XXY, XXXY, XXXXY, XYY and XXXYY. In general, any chromosome set carrying a Y chromosome will produce a male or at least male-like individual, though increasing numbers of X chromosomes in the above combinations generally lead to increasing "femaleness" in the male: development of breasts, widening of the pelvis, changes in pubertal hair

patterns, alteration of genitalia, etc. Conversely, persons lacking a Y chromosome are typically females (though atypical numbers of X are associated with varying degrees of sterility and mental dysfunction). What causes these unusual assortments of chromosomes? For experimental organisms, various precise answers are possible, including temperature, radiation, certain chemicals and advanced parental age. For humans, only advanced maternal age (increasing from age thirty-five on) has been reliably implicated and that for only some combinations.

There are also persons whose bodies are mosaics of chromosome constitutions—some cells containing one chromosome pattern, others containing another. These present a wide variety of combinations, even to a person with six separate types of cells: XXX/XXXY/XXXX/XXXXY/XXXXX/XXXXXY. Particularly interesting are those mosaics with combinations which opt for opposite sexual makeups: XX/XY, XX/XXY, XO/XY. Their physical characteristics will vary depending on a number of things; one of these is which specific body tissues are composed of each given chromosome combination. The group manifests a spectrum of body types, ranging from essentially normal females to essentially normal males. In between, of course, are those whose bodies are not clearly one sex or the other, but with the characteristics of both.

There is another significant category of individuals whose intersexual nature is unrelated to some unusual combination of chromosomes—those whose chromosomes appear to be numerically and structurally normal. Some knowledge of the development of the human embryo is important.

DEVELOPMENT OF SEX CHARACTERISTICS FROM EMBRYO TO PUBERTY

For the first several weeks of life, both sexes develop alike. The human embryo at the age of six weeks gives no anatomical evidence of which sex it will be. At this indeterminate stage, a series of structures common to both sexes has been produced. Even the primitive gonads, the "ovotestes," are each part female tissue (ovarian) and part male (testicular). Normally, one part of each gland will proliferate to form a functional gonad of the appropriate sex. But even under normal conditions, remnants of the "opposite" sex tissue remain in the gonad of both males and females.

Ordinarily, as the gonads develop they release hormones which trigger and coordinate the development of the related organs and external genitalia. This interplay of hormones is not simple because each sex normally releases low levels of the hormones characteristic of the opposite sex. The hormonal system of the brain is involved as well. Not only must the hormones be produced and released properly into the bloodstream, but the recipient cells of the genitalia must detect and respond to them at appropriate times and in precise ways. There are myriad points at which normal development may go awry and intersexes be produced. We cannot review all the known specific types; an examination of a few generalized ones will suffice.

First, there is a specific genetic condition which converts XY embryos, normally destined to be males, into females. It is usually called *testicular*

feminization, or sometimes *androgen insensitivity*. Even when the testes form normally and release the usual masculinizing hormones, the cells which should form the remainder of the reproductive structures do not respond to these hormones. Without the masculinizing hormones, the embryo tends to produce a "female" baby. Externally, such babies usually look perfectly normal; they are considered girls, and are raised as girls. No one has any reason to label them otherwise. They usually come to medical attention when, in spite of often normal pubertal female development, they fail to menstruate. Examination usually reveals no uterus or fallopian tubes—and a pair of testes in the abdominal position where ovaries would ordinarily be. Despite the testes and the XY chromosome constitution, such persons almost invariably consider themselves females: they were raised that way, they marry that way and there is no legitimate reason to question that identification. Where the vagina is too underdeveloped for normal coital function, corrective surgery is performed, and by adopting children, these women become successful mothers.

Another genetic condition, *adrenogenital syndrome*, is in some ways the opposite of testicular feminization: it converts XX embryos into males, or into a wide variety of sexual expressions ranging from clear-cut maleness to unquestioned femaleness. (As an aside, even though the sexual identity is often frustratingly confusing, there is considerable evidence that these persons have higher intelligence than normal.) Babies born with this syndrome are somewhat a "family choice"; they can be raised as either males or females. Since the children are XX, the gonads are usually ovaries. However, due to the abnormal production of a particular body hormone, the embryo becomes to some degree masculinized. At birth the doctor can be presented with equivocal external genitalia: Does this baby possess a small penis, or a large clitoris? Is this a male urethra that is not fully closed, or labia minora abnormally fused? An imperfect scrotum, or imperfect labia majora? Words cannot convey the enigma of these cases, only photographs or actual observation can do that. (The works listed by Money, and Money and Ehrhardt contain excellent illustrations.)

Although doctors differ, there does seem to be a general rule of thumb: If there is sufficient penile tissue to form an essentially normal and functional penis, the child should be raised as a boy. If not, surgery should promote the femaleness. In most cases hormone therapy is necessary and desirable, regardless of the chosen sex, to promote more normal body formation. With sufficient surgery, proper hormonal therapy and conscientious treatment by parents and family, these persons can enjoy an essentially normal adult life, marrying and rearing children (adopted, if necessary). The critical point is that persons with this syndrome can be either males or females. The condition is famous for its incredible plasticity.

Some persons with adrenogenital syndrome are raised throughout childhood as one or the other sex (based on medical sex declaration at the time of birth), but during puberty shift to the opposite sex in both body conformation (though not a total shift of genitalia) and self-image. (See Money's article on "Matched Pairs.")

The foregoing syndromes have involved intersexuality in which at least the gonadal condition has been relatively clear, once internal investigation has been made. There are also cases of "true" intersexuality (or true hermaphroditism) in which a single individual possesses gonadal tissue of both sexes. Though rare, medical literature now chronicles several hundred such persons. For more than one hundred, adequate chromosomal analyses have been made. The majority possess normal-appearing XX or XY constitutions; the remainder are primarily mosaics, e.g., XX/XY. Some of the latter, evidence indicates, began life as two separate embryos, one XX and normally destined to become a normal female, the other XY and potentially male. But the two embryos fused, forming one person, a mosaic true hermaphrodite. The condition has several other causes also and is manifest in a wide variety of body types, from near-normal maleness to near-normal femaleness. The external genitalia and associated internal ductwork and gonads can come in almost every imaginable combination. Again, depending on the specific details, these persons can be reared as either males or females. Corrective surgery and hormone substitution therapy are used to bring a more harmonious expression of the desired sex.

SEX CHANGE DUE TO MEDICAL/PSYCHOLOGICAL TREATMENT

Beyond these naturally occurring phenomena, babies also have been inadvertently shifted from apparent normality to intersexuality by well-intended medical treatments. A few years ago, a particular hormone therapy was used in the treatment of mothers who had a history of miscarriages. Quite unexpectedly, the hormones (progestins) masculinized female fetuses. Usually only an enlarged clitoris resulted, but in rare instances, a complete and well-formed penis (and empty scrotum) were formed. These children possessed ovaries, and nearly all have been raised as girls. Beyond feminization of the genitalia, no further surgery was required. This well-intended but unfortunate hormonal treatment, short-term though it was, emphasizes the plasticity of these physical aspects of sexual differentiation.

Thus far this article has considered anatomical features. Critical but exceedingly complex developments involving the brain and personal self-image go far beyond the scope of this discussion. A host of data shows that the manner of rearing, and the family behavior and structure can affect and alter gender identity. Some of the most dramatic cases are those of identical male twins who express different gender identities: one male, one female (cf. Green). One specific illustration is particularly thought-provoking. At the age of seven months, a pair of identical male twins were circumcised. Through a mishap, on one of the boys the penile tissue was totally lost. For ten months the parents wrestled with this problem, then began a program aimed at a complete switch of gender, including a change of name, girl's clothing and hair style. At 21 months, surgery for feminization of the external genitals was completed, and the child has since been raised as a girl. Now, after 14 years, the child shows every evidence that the program has been successful, and that her gender identity is fully comparable to normal

females. This case is not unique. There are others on record, though the presence of an identical twin makes this one especially valuable for study.

Readers who wish to pursue the literature further would do well to begin with the paperback book by Money and Ehrhardt. Their file of case histories (primarily at Johns Hopkins Hospital and School of Medicine, the world's premier research and therapy unit for these conditions) is a gold mine of data. Green's book is a well-written introduction to the field.

Gender identity, thus, is produced by an interaction of many factors, including at least the following: gene and chromosomal makeup, response of the fetal gonad, fetal and pubertal hormonal milieu, specific development of body and genitals in the fetus and in puberty, possible brain dimorphism, one's own body image and the behavior of other persons toward the developing child. Are there other factors also?

FROM A THEOLOGICAL VIEWPOINT

Consider a testicularly-feminized "female," who would be male but for one anomalous gene among the 100,000 or so which comprise humans. Does this body house a male, or a female, spirit? Such persons possess Y chromosomes and testes, yet they consider themselves female; they marry as females, adopt children—and are sealed as females in the temple. What are the eternal implications? Some persons with "adrenogenital" syndrome have been raised male, and some female. They, too, can marry and participate in the sacred ordinances. Have we articulated a theology to embrace this reality?

Some commentators have suggested that such "accidents" do not occur among Mormons, an erroneous statement presumably designed to resolve a perceived paradox. In fact, in a church of four million there are undoubtedly hundreds of such cases. Conservative estimates of the incidence among the general populace of chromosomal abnormalities per live births are for XXY, 1/800 male births; for XYY, 1/700 males; for XXX, 1/1000 females; for XO, 1/3,000 females (over 90% of which are naturally—spontaneously—aborted). Reliable figures for the incidence of the gene-caused syndromes (testicular feminization, adrenogenital syndrome, and related examples) are virtually impossible to obtain, but it is defensible to conclude that the major intersex conditions collectively account for at least one in each 25,000 persons, with minor anomalies being considerably more frequent.

There are other significant questions inherent in this challenging corner of human experience. As Mormons, we tend to emphasize that the body is the servant of the mind, or at least that it should be; that the body should reflect the wishes and higher aspirations of the mind; that the mind, in turn, can be equated with the spirit. In recent years, medical science has acknowledged for the first time the real problems of persons whose bodies are identifiably one sex—with or without the physical or hormonal miscues identified above—but whose minds are that of the opposite sex. In these cases, the mind/body guidelines have often been reversed. The ecclesiastical counsel frequently given to such persons is that the *body*, not the mind, is the man-

ifestation of God's will, and that by some means they should subject their minds to the morphology of their bodies. Is this an appropriate expression of the mind/spirit/body trichotomy? How does this relate to cases where gonadal tissue and body morphology of *both* sexes are expressed? Do our answers deal with the range of expression in such cases as adrenogenital syndrome?

"Authoritative" statements on this subject from the presiding authorities of the Church are too few and too oblique to permit or to justify analytical review. One can, if one is so inclined, string together a few public utterances which, though not specific, may be made to reflect a certain impatience with the problem. But this would be an injustice, for specific private communications and handling of individual cases reveal a much more cautious and sensitive approach.

It is surpassingly difficult for those of us with no gender problems to empathize with those who possess them; nevertheless, a genuine Christ-like commitment demands that we learn to do so. A sensitive and informed counselling program will require the thoughtful fusion of an inspired theology with an increasing wealth of biological understanding,—which is, after all, only revelation through another channel.

NOTES

[1]*The Fugger News-Letters*, ed. Victor Von Klarwill, p. 242–43.

[2]I have chosen to use the word "intersex" to indicate those conditions where normal gender identity is thrown into confusion but which are usually considered somewhat neutral under certain social mores and to facilitate consideration of the issues in an objective and sensitive manner. As I use the term, it does not include transvestism or homosexuality (male or female).

SELECTED BIBLIOGRAPHY

Armstrong, C. M. and A. J. Marshall, ed. 1964. *Intersexuality in Vertebrates Including Man.* Academic Press, New York.

Dewhurst, C. J. and R. R. Gordon. 1969. *The Intersexual Disorders.* Bailleire, Tindall, and Cassell, London.

Green, R. 1974. *Sexual Identity Conflict in Children and Adults.* Basic Books, New York.

Levitan, M. and A. Montagu. 1971. *Textbook of Human Genetics.* Oxford University Press, New York.

Maccoby, E., ed. 1966. *The Development of Sex Differences.* Stanford University Press, Stanford.

Money, J. 1970. "Matched Pairs of Hermaphrodites: Behavioral Biology of Sexual Differentiation from Chromosomes to Gender Identity," *Engineering and Science* (Calif. Inst. of Technology, special issue: Biological Bases of Human Behavior) 33(6): 34–39.

Money, J. 1968. *Sex Errors of the Body: Dilemmas, Education, Counseling.* Johns Hopkins University Press, Baltimore.

Money, J. and A. A. Ehrhardt. 1972. *Man and Woman, Boy and Girl, The Differentiation and Dimorphism of Gender Identity from Conception to Maturity.* Johns Hopkins University Press, Baltimore.

Vallet, H. L. and J. H. Porter, eds. 1979. *Genetic Mechanisms of Sexual Development.* Academic Press, New York.