

Q&A: Differences of sex development 101—it's complicated

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Professor Vincent Harley. Credit: Hudson Institute of Medical Research

Boy or girl? It's not always as simple as that. Internationally renowned researcher into differences of sex development (DSDs), Professor Vincent Harley, introduces the topic and answers your questions—separating fact from fiction.

Recent controversies in the sporting world have brought the issue of

gender into the public eye, and there is a great deal of misinformation circulating on the topic.

Prof Harley's research has been significant in improving our understanding of DSDs; he has also worked with major sporting bodies, including the International Olympic Committee, to develop rules governing transgender and intersex athletic participation.

As a starting point: a person's sex can be defined by [sex chromosomes](#), testosterone levels or genitalia.....but not always.

What is the difference between sex, gender, intersex and trans?

Sex refers to the physical appearance you were born with.

Gender identity refers to a fundamental sense of belonging and self-identification of being female, male, both, or something other than female or male. It does not necessarily correspond to the sex assigned or presumed at birth.

Sexual orientation is the sex of the person to whom one is sexually attracted. It may include heterosexual, bisexual, homosexual, pan-sexual, or asexual.

Intersex is an umbrella term for people whose reproductive organs, sexual anatomy or chromosome patterns differ from the biological definition of male or female. We call these conditions differences of sex development, or DSDs.

There are many ways someone can be intersex as it involves a spectrum of reproductive characteristics. Being intersex is not the same as being

Trans, which is when a person does not identify with their biological sex. Typically, [transgender people](#) look male or female at birth, very rarely have a DSD condition, and identify as the sex opposite to their appearance at birth.

How common are intersex conditions?

An estimated 1.7% of babies are born with intersex conditions, although many variations are not apparent at birth. About 1 in 2,000 newborns has genital differences that involve a sex differentiation specialist being consulted. Others are identified later in life, sometimes not until puberty or adulthood.

What causes intersex conditions?

Intersex traits can vary considerably and have a complex range of causes. Most are random but some are inherited. Chromosome variations can happen randomly and spontaneously during conception due to egg cells or sperm cells.

This means a child may not have the typically male (XY) or female (XX) chromosomes and instead have a combination such as XXY or XYY. Some intersex people do fit into either the XX or XY category but develop female anatomy on the outside and male anatomy on the inside and vice versa. The cause is not determined.

What does an intersex person look like?

Intersex traits can relate to external genitalia, internal anatomy, hormones or chromosome combinations different from XY (male) and XX (female).

Differences of sex development or DSDs are genetic conditions where a person's X or Y sex chromosomes or their gonads or genitals are not typical, sometimes making it unclear if a newborn baby is a boy or girl. DSDs are complex genetically and clinically, with potentially hundreds of different causes. These are often referred to as intersex conditions.

Being intersex is a natural variation in humans, it does not mean there is something wrong.

Is an intersex person more male or female?

Some intersex people have both ovarian and testicular tissue, while others have atypical female or male genitalia, or male/female genitalia with variations in their internal organs and/or hormones. Some intersex conditions may be related to underlying concerns that might require medical care, but people who are intersex are generally as healthy as those who are not.

Do intersex women have physical advantages?

There is no good evidence to suggest that they do. For a start, the percentage of intersex people competing in top level sport is roughly the same as the percentage in the community as a whole.

Some intersex people have higher levels of testosterone than most women, but in most cases that does not mean they will have a physical advantage. In many of these cases, they produce more testosterone, but their bodies lack the ability to process it into its most potent form. The best research available so far also suggests that women with high [testosterone levels](#) show no evidence of increased strength, stamina or lean muscle mass.

What happens when a baby is born intersex?

Historically, [medical research](#) aimed to explain why people were intersex and how it could be "fixed." A baby with intersex traits was usually assigned male or female, which often resulted in surgery or medication. It is now more widely understood that such difference does not mean physical makeup needs to be changed and that the person born intersex should have the right to consent to surgery.

More people believe that those born with intersex traits should not face unnecessary surgery or [medical treatment](#) and should have the opportunity to decide later for themselves how they would like to identify—intersex, male, female or otherwise. Not all people with intersex conditions have genital differences and some do not know they are intersex until their body does not develop as expected at puberty, or when they want to have children.

Are intersex people more or less healthy than the rest of the population?

Unless they have a related medical condition, babies born with intersex conditions are natural and healthy. Their long-term medical outlook is no different from other children.

Some DSDs result in medical issues such as problems making or regulating hormones, development issues during puberty and possibly increased cancer risk due to undeveloped tissue.

Early surgery to assign a sex may affect hormone levels later, and those born without various sex organs may need hormone replacement therapy.

Having intersex traits can also affect fertility, however with advanced

techniques such as IVF, an intersex person's sperms or eggs can be extracted and stored for fertilization later. Some intersex people have ovaries, a uterus and a vagina, and may be able to become pregnant or to carry a pregnancy.

Tell us about your research in this area

I am a molecular geneticist who has spent the past 30 years investigating the basis of sex development and gender identity. I head the Sex Development Research Group at the Hudson Institute of Medical Research in Melbourne.

Our intersex research aims to identify genes linked to intersex conditions, and the science behind underlying [reproductive organ \(gonad\) formation](#) in the developing embryo. This research has the potential to improve the understanding of what it is to be intersex, establish [possible genetic links](#) and causes, and [improve diagnosis](#) and clinical management of related health outcomes such as cancer risk, infertility, and gender dysphoria.

My team has classified 48 different forms of DSD among children born with ambiguous genitalia, as well as 12 new genes which have improved the rate of successful genetic testing from 15% to 50%. Based on these findings, they have developed a tool in the form of a decision tree to help inform the decision-making of DSD clinicians and families faced with newborns with ambiguous genitalia.

Parents usually expect to be able to say they have had a boy or a girl, so what happens in DSD cases?

Boy or girl? Sometimes the answer is not straightforward. While some DSDs are mild, and decisions on potential treatment can wait until the

child is older, some are life-threatening. A quick diagnosis and action is ideal, but this can be problematic for both parents and doctors.

Hudson Institute researchers Professor Vincent Harley and co-workers, Dr. Nayla Leon and Dr. Alejandra Reyes, introduced a new clinical guide to assist with the difficult decisions that can arise when a child is born intersex. From a systematic survey of DSD cases, a single tool was produced, a decision-tree, to help streamline diagnosis.

Their work was [published](#) in *The Lancet Diabetes and Endocrinology* in 2019.

The step-by-step guide helps specialists involved in teams who encounter newborn babies with ambiguous genitalia. Through enabling faster diagnosis, the clinical guide improves decision-making and, therefore, the future quality of life for many children and their families.

Provided by Hudson Institute of Medical Research

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