



The Gender Centre Inc. Fact Sheet

Ambiguous Genitalia

Definition and Causes

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When gender is unclear at birth

We're born, someone looks at our genitals, and instantly we're categorised. "It's a boy!" "It's a girl!" And from that one announcement, many things are determined. Whether we're issued a blue or pink blanket. Our name. To some extent, what toys and games we'll likely be given. Our future roles in reproduction.

But some babies are born with ambiguous genitals – it's not clear from just looking what their gender really is. And how gender is determined in such babies tells a lot about what actually makes someone male or female.

What does it mean to have ambiguous genitalia?

Ambiguous genitals refer to the uncertain appearance of a baby's external sexual features. Sometimes a female foetus is born with ovaries but male-like external genitals (female pseudohermaphroditism). A male may be born with testicles (which have yet to descend from the pelvis) but with female-like external genitalia (male pseudohermaphroditism). Rarely, newborns may even have both ovaries and testicles and ambiguous genitals (true hermaphroditism).

In addition, there may be other congenital defects present in these newborns, such as hypospadias in males. This is a condition in which the urethral (urinary) opening is not in its normal position on the tip of a penis but is on the underside.

What causes ambiguous genitalia?

The causes of ambiguous genitals include hormonal imbalances, chromosomal abnormalities and abnormalities of tissues that develop into genitals.

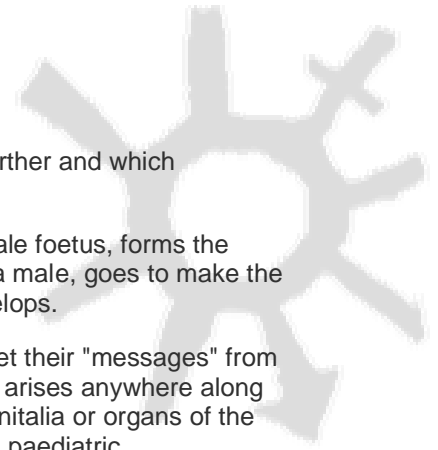
To understand how a human being could have both male and female sexual characteristics, one must understand the basics of sex differentiation.

At fertilisation, sperm and egg join and pool their chromosomes (genetic material), creating an embryo with 23 pairs. One of these 23 pairs is called the sex chromosomes. That pair is responsible for determining whether an embryo develops as a male or a female.

Almost all female embryos have two X chromosomes (XX). Almost all male embryos have an X and a Y chromosome (XY).

These sex chromosomes determine the differentiation of foetal gonads (rudimentary sex organs) into ovaries or testes. The hormones produced by the gonads (androgen and oestrogen) determine the differentiation of the external genitalia into male or female.

Early in foetal development, males and females are indistinguishable. Male foetuses have primitive male and female sex organs; female foetuses have primitive male and female sex



organs. It's the body's hormones that determine which organs develop further and which disappear.

As for external genitalia, the same foetal tissue that forms a penis in a male foetus, forms the clitoris in a female. And the same tissue that goes to make a scrotum in a male, goes to make the labia (lips of the vagina) in a female. Again, hormones dictate which develops.

"The hormones get their "messages" from the gonads, and the gonads get their "messages" from the sex chromosomes. So you can begin to understand how if a problem arises anywhere along these lines of "communication," a baby could be born with ambiguous genitalia or organs of the opposite gender," explains Donald Zimmerman, M.D. (Medical Doctor), a paediatric endocrinologist at Mayo Clinic, Rochester, Minnesota.

It's also important to understand that ambiguous genitalia are not the fault of the mother or father; in most cases, they cannot be detected or corrected pre-natally.

Two of the genetic abnormalities in foetuses that can lead to ambiguous genitalia are Androgen Insensitivity Syndrome (A.I.S.) and Congenital Adrenal Hyperplasia (C.A.H.).

A.I.S. is a condition in which foetal cells are unable to respond (to varying degrees) to the "male" hormone androgen. A newborn A.I.S. infant may have genitals of normal female appearance, but have a short vagina, no cervix or uterus and will have testes.

In C.A.H., an enzyme deficiency causes the adrenal glands to produce excess male hormones. C.A.H. can lead to enlargement of the penis in male infants and of the clitoris in female infants.

"Whatever the cause, what's important to understand is that ambiguous genitals are not an oddity of nature. These are simply parts of the body that are only part-way developed," Dr. Zimmerman explains. "It may be unclear at birth what was intended – a penis or clitoris. At that point, we have to look at several factors to determine gender."

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