

Intersex Conditions

Introduction

Intersex conditions are long-established genetic conditions that result in a child being born with sex chromosomes and/or sexual reproductive anatomy that are not exclusively male or female.

Intersex conditions include Androgen Insensitivity Syndrome (AIS), 5 alpha-reductase deficiency (5AR), 17-beta hydroxysteroid dehydrogenase deficiency (17B), XY gonadal dysgenesis (also known as Swyer Syndrome), Leydig cell hypoplasia, Mayer Rokitansky Kustner Hauser (MRKH) Syndrome (also known as vaginal agenesis), Congenital Adrenal Hypoplasia (CAH), Turner's Syndrome and Klinefelters Syndrome. Most people with these conditions are infertile and require life-long hormone treatment.

Types of intersex conditions

Androgen Insensitivity Syndrome.

People with AIS are genetically 46XY and are born with testes. Physical characteristics (phenotype) can vary from male to female and anywhere in between. People with Complete AIS (CAIS) are physically unambiguously female, whereas people with Partial AIS (PAIS) range from males with ambiguous genitals to women with little response to androgens. The condition is caused by an inability for foetuses with a male chromosomal pattern to respond either fully or partially to androgens (male hormones).

Congenital Adrenal Hypoplasia.

People with CAH are genetically 46XX and have female internal reproductive organs (vagina, uterus and ovaries). However, as a result of the adrenal glands of the developing foetus producing excessive amounts of androgen, the foetus' genitalia begin to develop in the male direction.

The clitoris of a baby with CAH is enlarged and may resemble a small penis. The cleft between the labia or lips may be partly closed over, hiding the entrance to the vagina. The urinary passage and vagina may also both open into one entrance as a result.

Hypospadias.

A condition where the urethral opening is not on the tip of the penis. (Usually along the penile shaft or in the perineum).

Gonadal Dysgenesis

A condition where an infant is born with underdeveloped gonads affecting foetal sexual development. Physical affects include sterility, genital appearance, osteoporosis and hormone deficiencies.

Klinefelters Syndrome

People with Klinefelter Syndrome are born with a 47XXY sex chromosome pattern. People with this condition are usually raised as males and often have breast development, low androgen production and small testes.

Turners Syndrome

A condition where an infant is born with a 45XO sex chromosome pattern. Typical affects include gonadal streaks in lieu of ovaries, short stature, cardiovascular

conditions and hormone deficiencies. Some physical manifestations of Turners syndrome have what is referred to as a Mosaic sex chromosome pattern where some or all cells in the body possess duplicate sex chromosomes such as XO/XY.

How common are these conditions?

Intersex Condition	Incidence
Klinefelter (47XXY)	1 in 1,000 births
Hypospadias	1 in 2,000 births
Androgen Insensitivity Syndrome (AIS)	1 in 13,000 births
Congenital Adrenal Hyperplasia (CAH)	1 in 13,000 births
Ovotestes	1 in 83,000 births
Partial Androgen Insensitivity Syndrome (PAIS)	1 in 130,000 births
Complete gonadal dysgenesis	1 in 150,000 births
5 alpha reductase deficiency	no estimate
Mixed gonadal dysgenesis	no estimate
'Ambiguous' genitalia	1 in 4,500 births

For more information:

AIS Support Group Australia

www.vicnet.net.au/~aissg