Patau's Syndrome Encyclopedia Article

Patau's Syndrome

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Contents

Patau's Syndrome Encyclopedia Article.....1

Contents.....2

Patau's Syndrome.....3

Patau's Syndrome

Patau's syndrome, also called trisomy 13, occurs when a child is born with three copies of chromosome 13. Normally, two copies of the chromosome are inherited, one from each parent. The extra chromosome causes numerous physical and mental abnormalities. Owing mostly to heart defects, the lifespan of trisomy 13 babies is usually measured in days. Survivors have profound mental retardation.

Individuals normally inherit 23 chromosomes from each parent, for a total of 46 chromosomes. However, genetic errors can occur before or after conception. In the case of Patau's syndrome, a random error occurs, and the embryo has three copies of chromosome 13, rather than the normal two copies.

Trisomy 13 occurs in approximately 1 in 12,000 live births. In many cases, spontaneous abortion (miscarriage) occurs, and the fetus does not survive. The risks of trisomy 13 seem to increase with the mother's age, particularly if she is over 30. Male and female children are equally affected, and the syndrome occurs in all races.

Newborns with trisomy 13 have numerous internal and external abnormalities. Commonly, the front of the brain fails to divide into lobes or hemispheres, and the entire brain is unusually small. Children who survive infancy usually exhibit profound mental retardation.

Incomplete development of the optic (sight) and olfactory (**smell**) nerves often accompanies the brain defects, and the child may also be deaf. Frequently, a child with trisomy 13 has cleft lip, cleft palate, or both. Facial features are flattened and ears are malformed and lowset. Extra fingers or toes may be present in addition to other hand and foot malformations.

Patau's syndrome can be detected during pregnancy through the use of ultrasonography, amniocentesis, and other testing. In infants the abnormality can be confirmed by examining the infant's chromosomal pattern. However, Patau's syndrome cannot be cured. Although certain structural abnormalities can be treated through surgery, malformations are often numerous and severe. Decisions regarding measures to prolong life are best made on an individual basis by parents and doctors. Medical treatment may simply focus on ameliorating symptoms rather than prolonging life.

Approximately 82% of trisomy 13 babies die within their first month of life; only 5-10% survive to one year. Children who survive infancy require medical treatment to correct structural abnormalities and associated complications. Survival to adulthood is very rare. Only one adult is known to have survived to age 33.