Patau Syndrome Encyclopedia Article

Patau Syndrome

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Patau Syndrome

Patau 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 baby 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 syndrome, an embryo develops which 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 older than her early 30s. Male and female children are equally affected, and the syndrome occurs in all races.

Patau syndrome is caused by the presence of three copies of chromosome 13. The presence of these three copies---rather then the normal two--is a random error and cannot be attributed to any action or lack of action on the part of the parents.

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 have 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 (polydactyly) may be present in addition to other hand and foot malformations.

In nearly all cases, trisomy 13 babies have respiratory difficulties and heart defects, including atrial and ventricular septal defects, patent ductus arteriosus, and defects of the pulmonary and aortic valves. Other organ systems may also be affected. The organ defects are frequently severe and life-threatening.

A newborn's numerous malformations indicate a possible chromosomal abnormality. Trisomy 13 is confirmed by examining the infant's chromosomal pattern through karyotyping or another procedure. Trisomy 13 is detectable during pregnancy through the use of ultrasonography, **amniocentesis**, and **chorionic villus sampling**.