John Hilton Edwards first described the symptoms of the genetic disorder known as Trisomy 18—one of the most common forms of human trisomy, which occurs when cells have an extra copy of a chromosome, in 1960. Trisomy 18, also known as Edwards Syndrome, occurs approximately once per 6000 live births and is second in frequency only to Trisomy 21, or Down’s Syndrome, as an autosomal trisomy. Trisomy 18 causes substantial developmental problems in utero.

The presence of an extra copy of chromosome 18 is a genetic anomaly that arises during the production of sperm and egg cells in either meiosis I, or more commonly meiosis II. Trisomy 18 results from defects in the mother’s eggs in 90 percent of its cases; further, the incidence rate increases with maternal age. During meiosis I, diploid cells contain two copies of each chromosome—one of which is maternal and the other paternal. Each chromosome duplicates and divides into two daughter cells. In meiosis II, these daughter cells divide further to produce four total haploid cells, with each having one set of chromosomes. These are gametes, and in humans they either occur as sperm or egg, depending on the sex of the parent. When sperm and egg fuse during sexual reproduction, their genetic information is combined in a new cell, the zygote, which has forty-six chromosomes. In cases of Trisomy 18, the cells of the zygote possess an extra chromosome 18.

Most instances of Trisomy 18 are full trisomies, meaning that every cell in the child’s body has the extra chromosome 18. As a result, significant genetic defects, such as extremely small body size and severe physical deformities, usually result in the death of the embryo or fetus, 95 percent of developing embryos and fetuses die before birth. Infants who are born with Trisomy 18 Survive to an average age of 14.5 days, and 8.4 percent live longer than a year. The oldest recorded life span of a person with Trisomy 18 is twenty-seven years. Although Trisomy 18 does not affect individuals based on his or her race, 80 percent of cases occur in females.

Newborns with Trisomy 18 experience severe psychomotor and growth retardation. They usually possess head sizes that are significantly smaller than an average child of the same age and sex. Furthermore, they also have small eyes (microphthalmia), malformed ears, a small mouth (microstomia), an undersized jaw (micrognathia), clenched fingers, and other malformations. In addition to these visible physical abnormalities, other characteristics of Trisomy 18 include the presence of holes between the lower chambers of the heart (ventricular septal defects) and holes between the upper chambers of the heart (atrial septal defects), malfunctioning kidneys, and the dislocation of the esophagus from the stomach (esophageal atresia).

Amniocentesis and chorionic villus sampling are effective prenatal methods for diagnosing Trisomy 18. The mortality rates for those who have Trisomy 18 are high, and there are
few medical treatments. Support organizations for parents, such as the Trisomy 18 [5] Foundation, offer written advice with titles such as Considering Saying Goodbye Early. Parents who seek treatment are further counseled on a case-by-case basis.

Although most medical literature that refers to Trisomy 18 [5] discusses it as a disease linked to a full trisomy of chromosome 18, partial and mosaic trisomies of the chromosome also exist. These trisomies, which occur when only some cells possess the extra genetic material, account for only 5 percent of Trisomy 18 [5] cases. Additionally, a form of Trisomy 18 [5], known as translocation trisomy—defined by an extra chromosome's attachment to another, independent chromosome—occurs in 2 percent of cases. Translocation trisomy results when a segment of chromosome 18 is present in triplicate. Symptoms of both partial and mosaic Trisomy 18 [5] are less severe than cases of full Trisomy 18 [5], and the affected infants experience a wide spectrum of effects. Those with mosaic and translocation trisomy often have similar physical anomalies as those who have full trisomy, such as microcephaly [15], heart defects, and developmental delay. However, their intellectual capabilities span from profound retardation [14] to above-average intelligence.

Though the severity of the symptoms of Trisomy 18 [5] ranges from fatal to manageable, the disorder is currently incurable. As one of the most common forms of trisomy in humans [11] and one of the few that can occur and still result in a live birth, Trisomy 18 [5] and its effects on the developing child have been well-documented and studied.

Sources


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