

# Example of edwards syndrome research paper

[Technology](#), [Development](#)



## **Definition and Causation of Edward's Syndrome**

Edwards's syndrome is a condition and severe genetic disorder caused by a chromosomal defect. The syndrome occurs in approximately 1 out of every 6,000 live born infant (Trisomy 18 Foundation, 2010). A human being is supposed to have 46 chromosomes, of which, 23 are obtained from the mother while the other 23 are obtained from the father. With in mind that conception is usually the fusion of male and female gametes (sperm and ovum), in case of an error in the formation of any of the two gametes, there is a high prospect that the baby formed from such gametes will most likely have a deformed or a wrong number of chromosomes. In this regard, Edwards's syndrome occurs when a baby, due to some defects of the sperm or the ovum, ends up having more than two copies of chromosome 18 in lieu of the normal two copies as shown in figure 1 below (Trisomy 18 Foundation, 2010). The syndrome can also occur when part of the chromosome 18 do not develop properly.

## **Characteristics of Edwards's syndrome**

With Edwards syndrome, are infants usually born small in size which is a pattern of physical features that in most cases serve as the cues used by health practitioners for the diagnosis of the disorder. The physical features associated with Edward's syndrome are inclusive of; elongated skull, small jaw, small mouth, malformed ear, short fissures of the eyelid, large ear lobes, and small fingernails among others (Support Organization for Trisomy 18, 13 and Related Disorders, 2012).

Also, the congenital malformations involve the presence of some internal or external birth defect, the most prominent is a defect of the heart which include: Ventricular septal defect (VSD) (in which there is a hole between the ventricles), Atrial septal defect (ASD) (which is a hole between the auricles) and Patent ducts arteriosis (PDA) (failure of the ductus arteriosus to close during childhood) (Trisomy 18 Foundation, 2010). Additionally, the most typical characteristics of the syndrome include: kidney problems, part of the intestinal tract, specifically the oesophagus, does not develop in a proper way (esophageal atresia), Excess amniotic fluid (polyhydramnios), Pocket of fluid on the brain (choroid plexus cysts), developmental delay, retardation in growth and Umbilical or inguinal hernia (Trisomy 18 Foundation, 2010).

## **Developmental implications**

Veritably, most children born with Edward's syndrome do not always live past the first year of life. Several research findings point to the fact that the reduced chances of growing past the one-year-old mark by children with Edwards syndrome is as result of a number of contributing factors. According to Support Organization for Trisomy 18, 13 and Related Disorders (SOFT) (2012) these contributing factors encompass; short cessation of in breathing, feeding problem, and prevalence of aspiration pneumonia. Other contributing factors to the deaths of victims of Edward's syndrome at an early age include; underdevelopment of the lungs and heart defects. It should be noted that even though heart defects can play some role in the causation of deaths of children with Edward's syndrome it can be proven that exceedingly few deaths are related to heart defects. The 5-10 percent of

children who survive their first year usually experience severe developmental disabilities, feeding difficulties, apnea, seizures, and urinary tract infections and require constant care in a bid to prevent or treat various complications associated with the syndrome (SOFT, 2012).

## References

18 Foundation. (n. d.). Trisomy 18 Foundation . What Is Trisomy 18?.

Retrieved October 8, 2012, from [http://www.trisomy18.org/site/PageServer?pagename=whatisT18\\_whatis](http://www.trisomy18.org/site/PageServer?pagename=whatisT18_whatis)

Organization for Trisomy 18, 13 and Related Disorders. (2012). Trisomy 18 Facts. Support Organization for Trisomy 18, 13 and Related Disorders.

Retrieved from <http://www.trisomy.org/wp-content/uploads/2012/09/Trisomy-18-Facts-8-20-12.pdf>