Edwards Syndrome (Trisomy 18) Fact Sheet

Edwards syndrome due to trisomy 18 is a medically complex condition of human chromosomes that occurs in approximately 1 in 6,000 live born infants. It is the second most common trisomy syndrome after Down syndrome/trisomy 21. Syndrome refers to a recognizable pattern of physical findings. Trisomy refers to three copies of a chromosome instead of the normal two. Therefore in Edwards syndrome there is a presence of an extra 18th chromosome, trisomy 18.

There are 3 types of chromosome findings found in persons with Edwards syndrome. About 95% of infants will have a **full or complete trisomy** in all body cells. The remainder 5% will have a **partial trisomy** due to a rearrangement of part or all of one chromosome (usually attached to another chromosome) called a **translocation**, or have **mosaicism** which is a mixture of two different cell lines (usually some normal cells and some trisomy 18 cells).

Most often a diagnosis is suspected by the results of maternal blood screening like noninvasive prenatal screening (NIPS) and fetal ultrasound findings. Diagnostic testing such as amniocentesis or chorionic villus sampling can be performed during the pregnancy to confirm a diagnosis. Otherwise testing can be done after the infant is born.

Three different categories of challenges are summarized below: birth defects and other conditions, child development, and survival.

Birth Defects

Almost all children born with Edwards syndrome will have birth defects; however each child is different, the occurrence is highly variable, and no child has all of these findings.

Cardiac (90% of infants)
- Ventricular septal defect (VSD)
- Atrial septal defect (ASD)
- Patent ductus arteriosus (PDA)
- Tetralogy of Fallot
- Valve disease

Respiratory
- Obstructive apnea
- Pulmonary Hypertension
- Tracheal-esophageal fistulas with esophageal atresia
- Diaphragmatic hernia
- Lung hypoplasia

Other
- Small at birth
- Spina Bifida
- Cleft lip with or without cleft palate
- Small mouth or lower jaw
- Clenched hands with index finger overlapping the 3rd digit
- Club feet

Common Disorders/Illnesses

- Small size compared to individuals of the same age
- Developmental delays
- Feeding difficulties
- Gastroesophageal reflux
- Chronic constipation
- Hearing loss
- Vision problems
- Strabismus
- Seizures
- Scoliosis
- Kidney defects
- Urinary tract infections
- Pulmonary hypertension
- Obstructive sleep apnea (OSA)

Child Development

Children will have developmental delays meaning that they will meet younger developmental milestones at older ages. Positive support, quality educational programs, stimulating home environment, and supportive therapies lead to increased enrichment of the children's lives. This leads to a wide range of developmental skills in language and communication in addition to gross and fine motor skills.

Survival

Survival in infants, children, and adults is constantly changing and improving. In the past it was said that only 6% would survive past 1 year. Recent studies now report increased survival in trisomy 18 especially with medical interventions; respiratory, nutritional, cardiac, and other surgeries when appropriate. The largest trisomy 18 survival study in the US reported trisomy 18 survival of 13.5% at 1 year, and 12.3% at 5 years. [Meyer et al., 2016] This however doesn't account for the survivals of 35-45% at 1 year if a child survives 1 month.

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