

Chromosomal Abnormalities: Trisomy

Overview:

Trisomy is a condition following the error of cell division in the formation of a gamete and results in three copies of one chromosome in cells that have two copies of the other chromosomes.

While trisomy of most chromosomes is lethal in humans, trisomy of chromosome 21 (the smallest chromosome) results in Down Syndrome.

Trisomy of a chromosome is indicated by the name of the chromosome affected. For example, trisomies that occur most often are listed below:

- Trisomy 8
- Trisomy 9
- Trisomy 12
- Trisomy 13
- Trisomy 18
- Trisomy 21

Incidence:

Apart from trisomy 21 (Down Syndrome), trisomies 13 and 18 are the only live born trisomies.

- **Trisomy 18**
 - Incidence of between 1:3,000 and 1:8,000 with a 3:1 Female: Male ratio.
 - Characteristics:
 - Clenched hand with overlapping fingers
 - Rocker bottom feet
 - Low set ears
 - Cardiac defects
 - Generalized muscle spasticity
 - Mental retardation
 - Mean survival time= 14.5 days
- **Trisomy 13**
 - Incidence of between 1:2,000 to 1:5,000 with an equal distribution of affected males and females.
 - Characteristics:
 - Seizures
 - Deafness
 - Microcephaly
 - Midline cleft lip & palate

- Abnormal ears
- Cardiac abnormalities
- Mental retardation
- Mean survival time= 7 days

Treatment:

While there has been an increase in the number of families who decide to continue with a pregnancy after a diagnosis of trisomy 13 or 18, the majority of such pregnancies (98%) have traditionally been terminated.

Research suggests that a highly individualized approach should be taken with a family who has been diagnosed with a pregnancy carrying trisomy 13 or 18. Such interdisciplinary approaches include determining the desired level of prenatal care, postpartum interventions, and genetic counseling.

Educational Concerns:

As 85% and 90% of children born with trisomy 13 and 18 (respectively) die by their first birthday, the role of the school psychologist with these children will be limited.

However, with the increasing number of families who choose to continue with known trisomy pregnancies, school psychologists have the opportunity to work with siblings of children carrying this difficult diagnosis.

Prolonged parental stress throughout the duration of the pregnancy will likely impact the sibling's academic, social, and emotional state in school. In addition, grief counseling with children will take a unique form as the child and family anticipate a birth associated with mixed emotions. Talking with children regarding expectations (physical abnormalities, short life-span) surrounding the birth of a special-needs sibling will help with fear of unknown or unexpected experiences in the hospital. Extra support for a sibling is likely needed when first informed of the diagnosis/prognosis of a trisomy pregnancy and in the time leading up to and immediately following birth and death.

Resources:

For more information:

<http://www.lpch.org/DiseaseHealthInfo/HealthLibrary/genetics/trisomy.html>

For families: <http://www.trisomy.org/>

For support: <http://www.hopefortrisomy13and18.org/>