THE CHROMOSOME 18 REGISTRY & RESEARCH SOCIETY

Trisomy 18p

Sixty-Second Summary

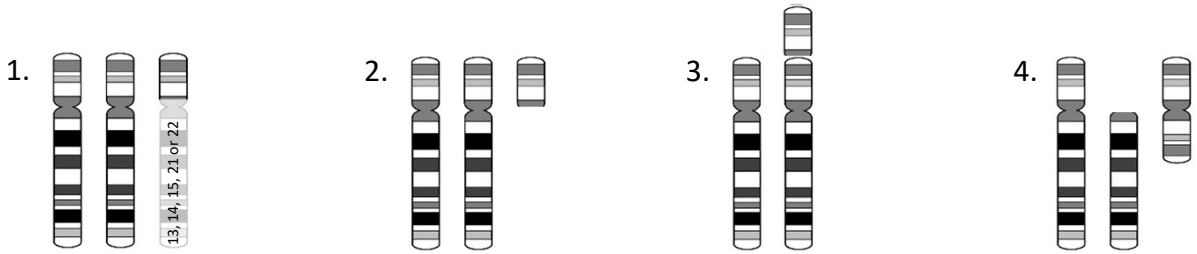
(Aliases: partial trisomy 18)

ICD-10 = Q93.2

UT Health San Antonio
The Chromosome 18 Clinical Research Center

Trisomy 18p

This a rare condition caused by 3 copies of the short arm (p arm)of chromosome 18. This results in a total of 3 copies of each of the 67 genes on 18p. Little is known about how the presence of three copies of these genes directly relates to the clinical features associated with Trisomy 18p. The information here is based on cases in the medical literature as well as from our own study participants.



Key points on genotype

- There are four chromosome configurations that can lead to Trisomy 18p
    1. An extra short arm can be translocated to another chromosome usually an acrocentric chromosome (Chromosome 13, 14 15 or 21)This occurs in 33% of cases.
    2. An extra short arm can be a supernumerary chromosome sometimes called a “marker chromosome.” This occurs in 30% of cases.
    3. An extra short arm can be duplicated in a tandem or inverted orientation in 26% of cases.
    4. The co-occurrence of two separate chromosome changes; an 18p deletion of one chromosome and an isochomosome 18p composed of 2 p arms. The combined result is Trisomy 18p and occurs in 11% of cases.
- 41% of the cases in the literature were of individuals whose diagnosis was incidental and subsequent to another family member’s diagnosis. They had not come to medical attention on their own.
- The familial cases were mothers with the same chromosome change or a parent with a balanced translocation.
- Parents may consider chromosome analysis to better define risks for future pregnancies

Key Points on phenotype

- Psychomotor delay is uncommon
- Mild cognitive delay is common
- Life expectancy is believed to be near normal
- Congenital anomalies are uncommon

Follow-up

- Recommendations for specific evaluations and treatments are in the following sections

Enrollment

- The Chromosome 18 Clinical Research Center is enrolling anyone with any chromosome 18 abnormality in our longitudinal study of all aspects of the conditions
- Parents may contact Annice Hill at hilla3@uthscsa.edu or call (210) 567-5321
- Enrollment requires the diagnostic genetics report and any other informative medical records

Consultation

- Daniel Hale, MD, Medical Director of the Chromosome 18 Clinical Research Center can be reached through Annice Hill at hilla3@uthscsa.edu or call (210) 567-5321

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