

Tetrasomy 18p

Sixty-Second Summary

(Aliases: isochromosome 18p)

ICD-10 = Q93.2



Tetrasomy 18p

This condition is caused by an extra chromosome composed of 2 chromosome 18 p arms (i.e. an isochromosome). This results in a total of 4 copies of each of the genes on 18p. The p arm of chromosome 18 has 67 genes. Little is known about how the presence of four copies of these genes directly relates to the clinical features associated with Tetrasomy 18p. Therefore, we base our recommendations on descriptive studies of individuals with this condition.

Key points on genotype

- Nearly all individuals with Tetrasomy 18p have the same genotype.
- Definitive diagnosis requires:
 - A chromosome microarray to determine the precise region of net copy number change.
 - A karyotype to demonstrate that the copy number change is due to an isochromosome.
- Almost all are *de novo* events as opposed to inherited from a parent.
- There have been a few case reports of parents with mosaicism or with a chromosome rearrangement.
- Parents may consider chromosome analysis to better define risks for future pregnancies.

Key Points on phenotype

- Most individuals are not medically fragile
- Developmental delay is very common
- The average full scale IQ score is 48, though there is a wide range of ability
- Behavioral concerns are common.
- Life expectancy is believed to be near normal.
- Congenital anomalies are possible. Specifically, individuals with Tetrasomy 18p have an increased likelihood of myelomeningocele, heart defects, hernias, palate abnormalities and orthopedic abnormalities.

Follow-up

- Affected individuals do not appear to be at increased risk for adverse reactions to drugs or standard medical treatments
- 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.