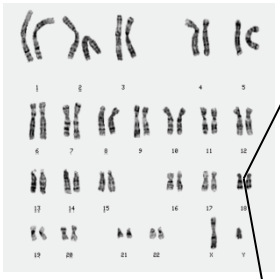
Ring 18

Sixty Second Summary

ICD-10 = Q99.9 or Q93.89



18p-
A deletion of any or all of the short arm. 18p has 67 genes.

Ring chromosomes involve a deletion of DNA from both ends of the chromosome, but not necessarily a deletion of genes from both ends.

Distal 18q- (18q21.1–q23)
A terminal deletion between 46,700,000 and the end of the chromosome at 78,077,248 bp.* The region includes 103 genes.

*hg 19 nucleotide scale

Key points on genotype

- Molecular and cytogenetic evaluations are both necessary to make a Ring 18 diagnosis.
- Every patient has unique deletions from BOTH ends of the chromosome; thus Ring 18 is not a single "syndrome." (For guidance developing an individualized gene-based interpretation see subsequent pages.)
- Some patients have deletions that do not include the deletion of any genes from one of the chromosome arms.
- Therefore, the clinical implication information is separated into 18q (Distal 18q-) and 18p (18p-) in the following pages.
- Some genotype-phenotype correlations have been established and are explored more in the following pages.
- ~20% have duplications just proximal to the breakpoint of up to 30 Mb in size
- Ring 18 occurs in 1 out of 300,000 live births

Key Points on phenotype

- Multiple congenital anomalies are possible. Specific phenotypes are dependent on the specific genes deleted - see the section of this report on molecular implications.
- Developmental delay is always present
- Intellectual disability is common but not inevitable
- Failure to thrive and growth hormone deficiency are common
- The risk for autism spectrum is higher than average
- Life expectancy is believed to be near normal except for individuals with very large duplications in addition to the deletions.

Management

- Affected individuals are not at increased risk for adverse reactions to drugs or standard medical treatments
- Treatment is primarily symptomatic
- 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 need to contact Annice Hill at hilla3@uthscsa.edu or call (210) 567-5321
- We need the diagnostic genetics report and any other informative medical records
- Then we will schedule a blood draw and shipment

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.