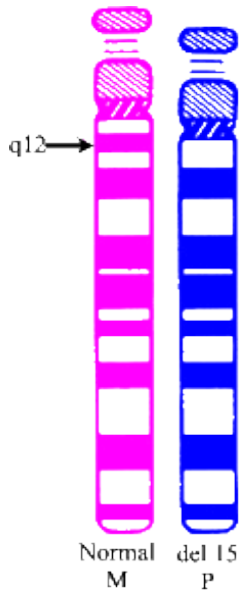




Genetics - Chromosome 15

P=Paternal
M=Maternal

An individual without Prader-Willi syndrome receives a chromosome 15 from the mother and another from the father. In Prader-Willi syndrome there are three genetic subtypes: deletion, maternal disomy and imprinting defect.



Deletion - 70%

A deletion including the q12 band (arrow) of chromosome 15 is contributed by the father and observed in about 70 percent of the subjects with Prader-Willi syndrome. The imprinted 15q11-q13 chromosome region includes genetic material (DNA) that is normally active (paternally expressed) on the father's chromosome 15 and inactive on the chromosome 15 inherited from the mother. Therefore, the paternally expressed genes from this region of chromosome 15 are missing in the deleted region of chromosome 15 and Prader-Willi syndrome occurs.

Laboratory Testing...

This typical common deletion of chromosome 15 can be seen at the microscope level and detected with high resolution techniques. The deletion is confirmed by fluorescence in situ hybridization (FISH) using DNA probes from chromosome 15.

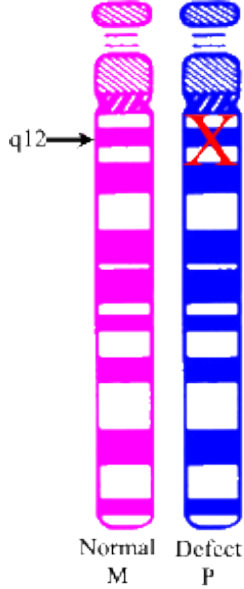


Maternal Disomy or UPD - 25%

In approximately 25 percent of subjects with Prader-Willi syndrome, both chromosome 15s are inherited from the mother (maternal disomy or UPD) and no chromosome 15 is present from the father. The missing chromosome 15 from the father which contains the active or paternally expressed genes required for normal development causes Prader-Willi syndrome in individuals in which both chromosome 15s are inherited from the mother.

Laboratory Testing...

Genetic testing is required to identify maternal disomy 15 or UPD using DNA obtained from the parents and the person with Prader-Willi syndrome. Polymorphic DNA markers from chromosome 15 are studied to identify whether both chromosome 15s are from the mother.



Imprinting Defect - 5%

A few individuals with Prader-Willi syndrome (up to 5 percent) may have an imprinting defect inherited from the father of the chromosome 15 involving the q11-q13 region. This imprinting defect will not allow the normal expression of genes that are active on the chromosome 15 from the father and needed for normal development. Prader-Willi syndrome then occurs. Rarely these imprinting defects on the father's chromosome 15 may be inherited with a potential 50 percent risk for having additional children with Prader-Willi syndrome.

Laboratory Testing...

To identify imprinting defects on chromosome 15 requires specialized laboratory techniques available in only a few genetic laboratories conducting research in Prader-Willi syndrome.

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