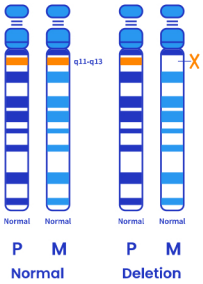
Genotypes of Angelman Syndrome



Deletion (65-75%)

DNA (deoxyribonucleic acid) is the main component of chromosomes. It contains our unique genetic code. Most individuals with AS are missing a piece of DNA in region 15q11-13 on the maternal chromosome 15.

Uniparental Disomy (UPD) (3-7%)

An individual with UPD has two copies of chromosome 15 from their father, instead of one each from the father and mother.

Mutation (5-11%)

This occurs when there is a small abnormality in the DNA of the *UBE3A* gene. A mutation can happen anywere on the gene.

Imprinting Center Defect (ICD) (<3%)

ICD occurs when there is an abnormality in the imprinting center of the mother's copy of the 15th chromosome. The imprinting center is in the area of the chromosome that controls whether genes are turned on or off. In ICD, the *UBE3A* gene from the mother is typically present, but an abnormality in the imprinter center prevents the mother's copy of the gene from being read properly in neurons as it is "turned off".