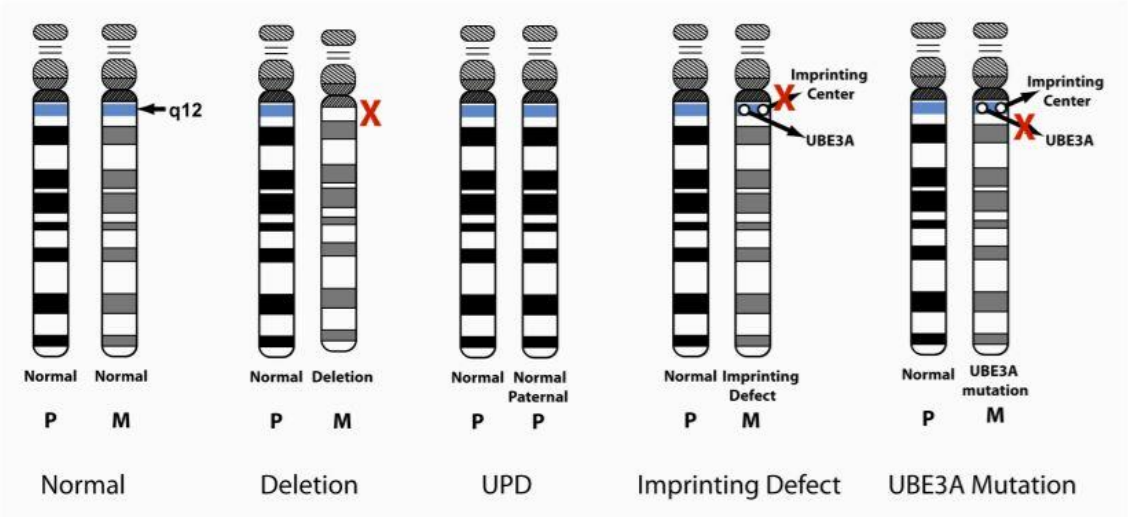
Genetic Classes of Angelman Syndrome



1. **Deletion+** (15q11.2-q13 deletions), 68% of cases – the majority of AS cases are caused by deletions on the maternal copy of Chromosome 15.
2. **UBE3A mutations**, 11% of cases – Mutations in the UBE3A gene either prevent its expression or function, thus these individuals do not have the appropriate levels of functional UBE3A in the brain.
3. **Uniparental disomy**, UPD; 7% of cases –The individual has two copies of paternal Chromosome 15.
4. **Imprinting defect**, ICD, 3% of cases – A deletion of the imprinting centre on Chromosome 15. Could also be caused by loss of imprinting information during the mother's oogenesis.
5. **Clinical/other**, 11% – When all testing for Angelman Syndrome is negative but they still meet the diagnostic criteria for AS. These individuals may have as yet unrecognized mutations that affect UBE3A or genomic imprinting on Chromosome 15.