# MED12 Related Disorders

*https://pubmed.ncbi.nlm.nih.gov/24123922/*

MED12: is a member of the large Mediator complex, which has a critical and central role in RNA polymerase II transcription. As a multiprotien complex, Mediator regulates signals involved in cell growth, development, and differentiation, and it is involved in a protein network required for extraneuronal gene silencing and also functions as a direct suppressor of Gli3-dependent Sonic hedgehog signaling. This may explain its role in several different X-linked intellectual disability syndromes that share some overlapping clinical features. This review will compare and contrast four different clinical conditions that have been associated with different mutations in MED12, which is located at Xq13. To date, these conditions include Opitz-Kaveggia (FG) syndrome, Lujan syndrome, Ohdo syndrome (Maat-Kievit-Brunner type, or OSMKB), and one large family with profound X-linked intellectual disability due to a novel c.5898insC frameshift mutation that unlike the other three syndromes, resulted in affected female carriers and truncation of the MED12 protein. It is likely that more MED12 mutations will be detected in sporadic patients and X-linked families with intellectual disability and dysmorphic features as exome sequencing becomes more commonly utilized, and this overview of MED12-related disorders may help to correlate MED12 genotypes with clinical findings.