

ATR-X

<https://pubmed.ncbi.nlm.nih.gov/11449489/>

Since the identification of the ATRX gene (synonyms XNP, XH2) in 1995, it has been shown to be the disease gene for numerous forms of syndromal X-linked mental retardation [X-linked alpha thalassemia/mental retardation (ATR-X) syndrome, Carpenter syndrome, Juberg-Marsidi syndrome, Smith-Fineman-Myers syndrome, X-linked mental retardation with spastic paraplegia]. An attempt is made in this article to review the clinical spectrum associated with ATRX mutations and to analyse the evidence for any genotype/phenotype correlation.