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Alpha Thalassemia X-Linked Mental Retardation Syndrome

National Cancer Institute

Source

National Cancer Institute. *Alpha Thalassemia X-Linked Mental Retardation Syndrome*.
NCI Thesaurus. Code C118631.

A rare, X-linked recessive inherited syndrome caused by mutations in the ATRX gene. It is characterized by intellectual disability, developmental delays, hypotonia, widely spaced eyes, small nose, low-set ears, tented upper lip, skeletal abnormalities, and a mild form of alpha thalassemia.