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Alpha-thalassemia-X-linked intellectual disability syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Alpha-thalassemia-X-linked intellectual disability syndrome. ORPHA:847

X-linked alpha thalassaemia mental retardation (ATR-X) syndrome in males is associated with profound developmental delay, facial dysmorphism, genital abnormalities and alpha thalassaemia. Female carriers are usually physically and intellectually normal.