X-linked creatine transporter deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. X-linked creatine transporter deficiency. ORPHA:52503

X-linked creatine transporter deficiency (CRTR-D) is a creatine deficiency syndrome characterized clinically by global developmental delay/ intellectual disability (DD/ID) with prominent speech/language delay, autistic behavior and seizures.