DISEASE:
X-linked creatine transporter deficiency

NAME:	X-linked creatine transporter deficiency
DESCRIPTION:	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.
ORPHACODE:	52503
SYNONYMS:	Creatine transporter deficiency SLC6A8 deficiency
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	SLC6A8
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