

DISEASE:

Global developmental delay-neuro-ophthalmological abnormalities-seizures-intellectual disability syndrome

NAME:	Global developmental delay-neuro-ophthalmological abnormalities-seizures-intellectual disability syndrome
DESCRIPTION:	A rare genetic neurological disorder characterized by infantile to childhood onset of global developmental delay, hypotonia, seizures, growth delay, and intellectual disability. Additional variable features include strabismus, cortical visual impairment, nystagmus, movement disorder (such as dystonia, ataxia, or chorea), or mild dysmorphic features, among others.
ORPHACODE:	488613
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	GNB1
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