DISEASE:
Xq25 microduplication syndrome

NAME:	Xq25 microduplication syndrome
DESCRIPTION:	A rare, X-linked, multiple congenital anomalies/dysmorphic malformation-intellectual disability syndrome characterized by developmental delay, mild to moderate intellectual disability, speech disturbance, behavioral problems (such as anxiety, hyperactivity, and aggressiveness) and mild facial dysmorphism (including facial hypotonia, thin arched eyebrows, ectropion, epicanthus, malar flatness, thick vermillion of the lips and prognathia). Additional variable manifestations include short stature, skeletal and genital anomalies, seizures, and autism spectrum disorders. Brain imaging may reveal cerebellar vermis hypoplasia, thin corpus callosum, and enlarged subarachnoid spaces.
ORPHACODE:	521258
SYNONYMS:	Dup(X)(q25) Xq25 microtriplication
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	STAG2
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- STAG2 cohesin complex component

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