
DISEASE:
Down syndrome

NAME:	Down syndrome
DESCRIPTION:	Down syndrome is a chromosomal abnormality caused by the presence of a third (partial or total) copy of chromosome 21 and that is characterized by variable intellectual disability, muscular hypotonia, and joint laxity, often associated with a characteristic facial dysmorphism and various anomalies such as cardiac, gastrointestinal, or endocrine defects.
ORPHACODE:	870
SYNONYMS:	Trisomy 21
XREF(S):	<u>Orphanet</u>
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