DISEASE:
Weaver syndrome

NAME:	Weaver syndrome
DESCRIPTION:	Weaver syndrome (WVS) is a rare, multisystem disorder characterized by tall stature, a typical facial appearance (hypertelorism, retrognathia) and variable intellectual disability. Additional features may include camptodactyly, soft doughy skin, umbilical hernia, and a low hoarse cry.
ORPHACODE:	3447
SYNONYMS:	Camptodactyly-overgrowth-unusual facies syndrome
XREF(S):	Orphanet OMIM ICD-10 OMIM MeSH OMIM
ANALYTE(S):	EED NSD1 EZH2 SUZ12
CREATED:	13 May 2019 - 01:02

CHANGED:	22 Jun 2023 - 16:14

Source URL: http://gentest.healthdata.be/disease/365

RELATED CONTENT

Related Genetic Tests

- Overgrowth (gene panel)

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)

Related Analytes

- embryonic ectoderm development
- enhancer of zeste 2 polycomb repressive complex 2 subunit
- nuclear receptor binding SET domain protein 1
- SUZ12 polycomb repressive complex 2 subunit

Related Gene Panels

- Overgrowth (24 genes) - IPG