DISEASE:
Wiedemann-Steiner syndrome

NAME:	Wiedemann-Steiner syndrome
DESCRIPTION:	A rare, genetic multiple congenital anomalies/dysmorphic syndrome characterized by short stature, hypertrichosis (most commonly of the back or elbow regions), facial dysmorphism, behavioral problems, developmental delay and, most commonly, mild to moderate intellectual disability.
ORPHACODE:	319182
SYNONYMS:	Hypertrichosis-short stature-facial dysmorphism-developmental delay syndrome
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	KMT2A
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Genetic Tests

- Short stature/ Growth retardation/ (gene panel)

Related Laboratories

- Centrum Medische Genetica - UZ Antwerpen

Related Analytes

- lysine methyltransferase 2A

Related Gene Panels

- Growth retardation/short stature (genepanel) - UZA