DISEASE:
Baraitser-Winter cerebrofrontofacial syndrome

NAME:	Baraitser-Winter cerebrofrontofacial syndrome
DESCRIPTION:	Baraitser-Winter syndrome (BWS) is a malformation syndrome, characterized by facial dysmorphism (hypertelorism with ptosis, broad bulbous nose, ridged metopic suture, arched eyebrows, progressive coarsening of the face), ocular coloboma, pachygyria and/or band heterotopias with antero-posterior gradient, progressive joint stiffening, and intellectual deficit of variable severity, often with severe epilepsy. Pachygyria - epilepsy - intellectual disability - dysmorphism (Fryns-Aftimos syndrome (FA); see this term) corresponds to the appearance of BWS in elderly patients.
ORPHACODE:	2995
XREF(S):	Orphanet OMIM OMIM ICD-10
ANALYTE(S):	ACTB ACTG1
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RELATED CONTENT

Related Genetic Tests

- Epilepsy (gene panel)
- Epilepsy, seizures (gene panel)
- Trombosis - Hemostasis (gene panel)

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Medische Genetica - UZ Antwerpen
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- actin beta
- actin gamma 1

Related Gene Panels

- Epilepsy, seizures (196 genes) - IPG
- Rare epilepsy with developmental delay (> 240 genes) - UZA
- Trombosis - Hemostasis (107 genes) - KUL