BWCFF Baraitser-Winter Cerebrofrontofacial Syndrome

https://pubmed.ncbi.nlm.nih.gov/26583190/

Clinical characteristics:

Baraitser-Winter cerebrofrontofacial (BWCFF) syndrome is a multiple congenital anomaly syndrome characterized by typical craniofacial features and intellectual disability. Many (but not all) affected individuals have pachygyria that is predominantly frontal, wasting of the shoulder girdle muscles, and sensory impairment due to iris or retinal coloboma and/or sensorineural deafness. Intellectual disability, which is common but variable, is related to the severity of the brain malformations. Seizures, congenital heart defects, renal malformations, and gastrointestinal dysfunction are also common.

Diagnosis/testing:

The diagnosis of BWCFF syndrome is established in a proband with suggestive findings and a heterozygous missense pathogenic variant in either

ACTB

or

ACTG1

identified by molecular genetic testing.

Management:

Treatment of manifestations:

Standard treatment for medical concerns in conjunction with the associated specialist; management of developmental delay and intellectual disability is tailored to the individual.

Surveillance:

Routine follow up recommended for neurodevelopmental assessment in all; follow up as needed for those with seizures (neurologic evaluation), coloboma or microphthalmia (ophthalmologic evaluation), hearing loss (audiologic evaluation), cardiac defects, renal tract anomalies, and gastrointestinal dysfunction.

Genetic counseling:

BWCFF syndrome is an autosomal dominant disorder. Most individuals with BWCFF syndrome reported to date have the disorder as the result of a

de novo

ACTB

or

ACTG1

pathogenic variant. If a parent of the proband has the pathogenic variant identified in the proband, the risk to the sibs of inheriting the pathogenic variant is 50%. Once the

ACTB

or

ACTG1

pathogenic variant has been identified in an affected family member, prenatal and preimplantation genetic testing for BWCFF syndrome are possible.