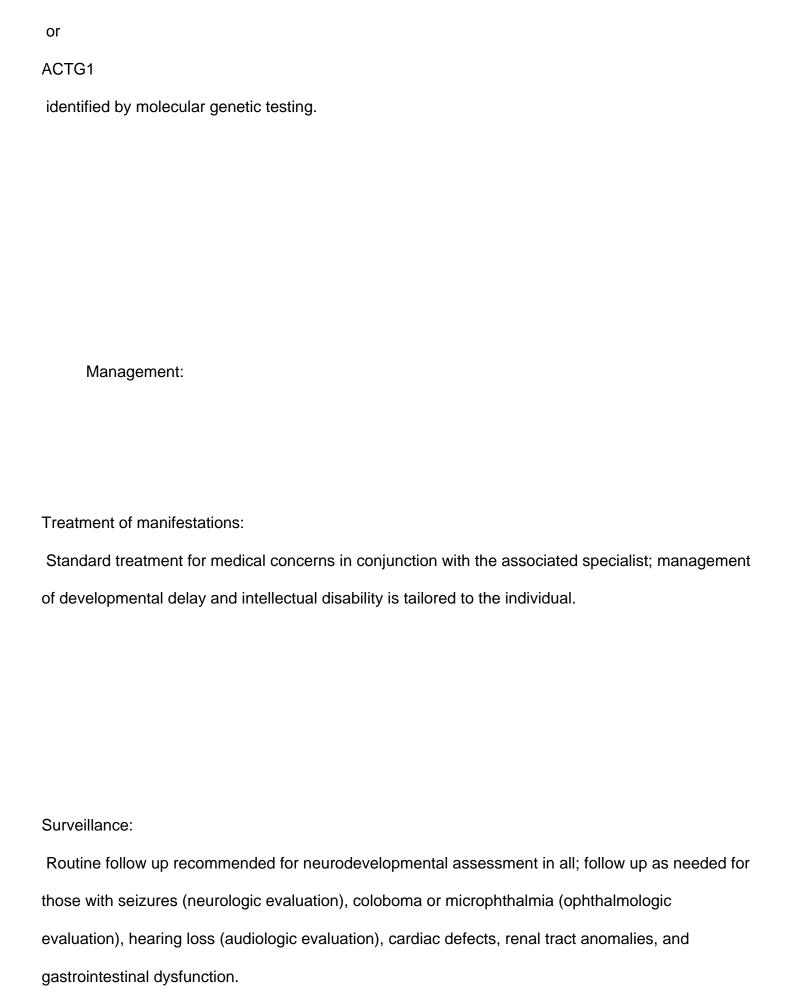
BWCFF Baraitser-Winter Cerebrofrontofacial Syndrome

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



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.