BWCFF Baraitser-Winter Cerebrofrontofacial Syndrome

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Baraitser-Winter cerebrofrontofacial syndrome (BWCFF) (BRWS; MIM #243310, 614583) is a rare developmental disorder affecting multiple organ systems. It is characterised by intellectual disability (mild to severe) and distinctive facial appearance (metopic ridging/trigonocephaly, bilateral ptosis, hypertelorism). The additional presence of cortical malformations (pachygyria/lissencephaly) and ocular colobomata are also suggestive of this syndrome. Other features include moderate short stature, contractures, congenital cardiac disease and genitourinary malformations. BWCFF is caused by missense mutations in the cytoplasmic beta- and gamma-actin genes ACTB and ACTG1. We provide an overview of the clinical characteristics (including some novel findings in four recently diagnosed patients), diagnosis, management, mutation spectrum and genetic counselling.