BWCFF Baraitser-Winter Cerebrofrontofacial Syndrome

https://pubmed.ncbi.nlm.nih.gov/27868373/

Baraitser-Winter malformation syndrome (BWMS), Fryns-Aftimos syndrome (FA), and craniofrontofacial syndromes (CFFs) have all been recently proposed to be part of the same phenotypic spectrum of Baraitser-Winter cerebrofrontofacial syndrome (BWCFF), which is characterized by facial dysmorphism, ocular coloboma, brain malformations, and intellectual disabilities. In addition to that, the recent discovery of missense mutations in one of the two ubiquitously expressed cytoplasmic β- and γ-acting-encoding genes ACTB (7p22.1) and ACTG1 (17q25.3) in patients carrying a clinical diagnosis of BWSM, FA, or CCF has provided further evidence that these clinical conditions do indeed belong to the same entity at the molecular level. Two cases of BWCFF patients presenting with malignancies (i.e., acute lymphocytic leukemia and cutaneous lymphoma) have been published thus far. Here, we report a 21-year-old female with molecularly confirmed FA, who developed acute myeloid leukemia (AML). The present finding may indicate that actinopathies could be cancer-predisposing syndromes although small numbers and publication bias should be taken into account. © 2016 Wiley Periodicals, Inc.