Baraitser-Winter cerebrofrontofacial syndrome: delineation of the spectrum in 42 cases

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Résumé en anglais	Baraitser-Winter, Fryns-Aftimos and cerebrofrontofacial syndrome types 1 and 3 have recently been associated with heterozygous gain-of-function mutations in one of the two ubiquitous cytoplasmic actin-encoding genes ACTB and ACTG1 that encode beta- and gamma-actins. We present detailed phenotypic descriptions and neuroimaging on 36 patients analyzed by our group and six cases from the literature with a molecularly proven actinopathy (9 ACTG1 and 33 ACTB). The major clinical anomalies are striking dysmorphic facial features with hypertelorism, broad nose with large tip and prominent root, congenital non-myopathic ptosis, ridged metopic suture and arched eyebrows. Iris or retinal coloboma is present in many cases, as is sensorineural deafness. Cleft lip and palate, hallux duplex, congenital heart defects and renal tract anomalies are seen in some cases. Microcephaly may develop with time. Nearly all patients with ACTG1 mutations, and around 60% of those with ACTB mutations have some degree of pachygyria with anteroposterior severity gradient, rarely lissencephaly or neuronal heterotopia. Reduction of shoulder girdle muscle bulk and progressive joint stiffness is common. Early muscular involvement, occasionally with congenital arthrogryposis, may be present. Progressive, severe dystonia was seen in one family. Intellectual disability and epilepsy are variable in severity and largely correlate with CNS anomalies. One patient developed acute lymphocytic leukemia, and another a cutaneous lymphoma, indicating that actinopathies may be cancer-predisposing disorders. Considering the multifaceted role of actins in cell physiology, we hypothesize that some clinical manifestations may be partially mutation specific. Baraitser-Winter cerebrofrontofacial syndrome is our suggested designation for this clinical entity.European Journal of Human Genetics advance online publication, 23 July 2014; doi:10.1038/ejhg.2014.95.
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