Syndrome of congenital cataracts, sensorineural deafness, Down syndrome-like facial appearance, short stature, and mental retardation: two additional cases

Abstract

An apparently new syndrome of congenital cataracts, sensorineural deafness, Down syndrome-like facial appearance, short stature, and mental retardation was described by Gripp et al. 1996. The authors reported on two unrelated patients with congenital cataracts, sensorineural deafness, distinctive facial appearance, mental retardation, postnatal short stature, and skeletal changes. We report on two additional patients with findings most similar to the reported patients by Gripp et al. 1996, including bilateral congenital cataracts, hearing loss, craniofacial abnormalities, short stature, skeletal abnormalities, and developmental delay. Both of the patients reported herein had chromosome microarray analysis, which showed normal results in Patient 2 but abnormal results in Patient 1 and his mother who both had a chromosome 11q25 subtelomere deletion. Patient 1 and his mother's findings are atypical for the common findings reported in Jacobsen syndrome (11q terminal deletion syndrome), and consistent with the patients reported by Gripp et al. 1996. The etiology for these cases has been unknown. The microarray results on Patient 1 suggest that the other patients with findings of developmental delay, short stature, congenital cataracts, sensorineural hearing loss, and similar craniofacial features may have either a microdeletion of

chromosome 11q terminal region or haploinsufficiency of a gene localized to this region.

Keppler-Noreuil K, Welch J, Baker-Lange K. Syndrome of congenital cataracts, sensorineural deafness, Down syndrome-like facial appearance, short stature, and mental retardation: two additional cases. Am J Med Genet A. 2007 Nov 1;143A(21):2581-7. doi: 10.1002/ajmg.a.31990. PMID: 17935251.