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Titel des Beitrags:
KIF21A variant R954W in familial or sporadic cases of CFEOM1.

Abstract:
PURPOSE: To demonstrate the clinical characteristics and determine mutations in the KIF21A gene, encoding a kinesin motor protein in patients with congenital fibrosis of the extraocular muscles (CFEOM) type 1. METHODS: Patients of five families with congenital fibrosis syndrome and two simplex patients with CFEOM underwent ophthalmologic examination and mutation analysis in the KIF21A gene. RESULTS: Clinical examination and passive motility testing prior to surgery met criteria for CFEOM. All patients had congenital restrictive ophthalmoplegia primarily affecting muscles innervated by the oculomotor nerve. Complete mutation screening in the KIF21A gene revealed the presence of the known and most common recurrent variant R954W in three families and in two simplex cases. Two families demonstrated linkage to chromosome 16. CONCLUSIONS: The patients included in the study had marked restriction of movement bilaterally with nearly complete loss of vertical ocular motility, graded reduction of horizontal motility, ptosis, and compensatory chin elevation. The phenotype was variable in patients carrying the same mutation. In one family, all patients were diagnosed with mental retardation, indicating that this syndrome might not only affect the development of cranial nerves, but can also be responsible for general neurologic dysfunction. The screening data suggest frequent and exclusive appearance of the R454W variant in
sporadic and familial cases of CFEOM1 in Germany.