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Graefe's Archive for Clinical and Experimental Ophthalmology
Publisher: Springer-Verlag Heidelberg
ISSN: 0721-832X (Paper) 1435-702X (Online)
DOI: 10.1007/s00417-003-0703-z
Issue: Volume 241, Number 7
Date: July 2003
Page: 546

Clinical Investigation

Congenital fibrosis syndrome associated with central nervous system abnormalities

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Received: 7 August 2002 **Revised:** 9 April 2003 **Accepted:** 7 May 2003 **Published online:** 18 June 2003

Abstract
Background Congenital fibrosis of extraocular muscles (CFEOM) is a complex strabismus syndrome that typically occurs in isolation and results from dysfunction of all or part of cranial nerves III (CNIII) and IV (CNIV) and/or the muscles that these nerves innervate. Only a few patients with CFEOM and additional central nervous system malformations have been reported. We describe four additional patients with CFEOM associated with central nervous system (CNS) abnormalities.
Methods Four patients who presented with congenital restriction of eye movements in association with neurological abnormalities underwent complete ophthalmological examination including electroretinography (ERG) and eye movement recordings. Neurological examinations, neuroradiological studies, muscle histology, chromosomal and genetic linkage analysis were performed.
Results Clinical examination and forced duction testing confirmed that all four patients met criteria for CFEOM; all had congenital restrictive ophthalmoplegia primarily affecting extraocular muscles innervated by the oculomotor nerve. Two brothers had CFEOM and Marcus Gunn jaw winking. In each of the four cases, CFEOM occurred in association with one or several neuroradiological findings, including agenesis of the corpus callosum, colpocephaly, hypoplasia of the cerebellar vermis, expansion of the ventricular system, pachygyria, encephalocele and/or hydrancephaly.
Conclusions We present four cases of CFEOM in association with CNS malformations that confirm that CFEOM can be part of a more complex neurological dysfunction and provide further support to a neurogenic aetiology for this disorder. We also describe for the first time the coexistence of CFEOM and Marcus Gunn jaw winking in two siblings. This suggests a genetic mechanism. Aberrant innervation supports primary developmental abnormality of cranial nerves in CFEOM.