Autosomal dominant optic atrophy with unilateral facial palsy: a new hereditary condition?

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Abstract
A mother and daughter are reported with bilateral optic atrophy with onset in infancy and unilateral facial palsy. This appears to be a novel autosomal dominant disorder.

Case reports
Case 1 is a female infant, the first of two sibs born to unrelated parents. She was born at term weighing 3960 g. Labour was induced because of postmaturity but the cervix remained unfavourable and a flat cardiotocograph trace led to emergency caesarean section. Delivery was aided by forceps.

On neonatal examination, she was found to have a right side facial palsy, which was initially thought to be a neuropraxia following the application of forceps. This was therefore treated expectantly but did not resolve.

Nystagmus was detected shortly after birth and led to the finding of optic atrophy. Full blood count, urea and electrolytes, serum amino acids, serum immunoglobulins, and TORCH screen were normal. CT scans and magnetic resonance brain scans were all normal; in particular, there was no evidence of any underlying neurodegenerative disorder or nerve compression.

At 4 years of age the girl is functioning as partially sighted with reduced visual acuity of 6/36+1 right, 6/36+2 left. Her right facial palsy persists. She has a good Bell’s phenomenon and, although the eye occasionally gets irritated, it responds well to lubricants. The rest of her central nervous system is normal and she has no other cranial nerve deficits. Her developmental milestones, hearing, and cognition are normal. She is of proportionate tall stature (height 116.5 cm and weight 26.5 kg, both greater than the 97th centile; maternal height 166 cm, 75th centile; and paternal height 93 cm, greater than the 97th centile). She is currently awaiting a facial nerve transplant to try to improve her appearance and symptoms.

Case 2 is the mother of case 1. She was an only child and is registered as partially sighted with bilateral optic atrophy. She presented in infancy with nystagmus and her mother was informed of her poor vision at about 4 months of age. She had attended a school for visually impaired children from the age of 11 years. Her visual impairment appears to have been non-progressive. She is of normal intelligence and normal hearing.

Close examination of the mother shows that she too has a slight right facial palsy (fig 1). Her face is asymmetrical; the right nasolabial fold is less pronounced than the left. Eye closure is full but the periorbital muscles on palpation are appreciably weaker than on the left. Photos of her as an infant and toddler show the facial palsy was present then.

There is no other family history of either optic atrophy or facial palsy. Case 1’s younger...
brother appears to be developing normal vision and has no nystagmus.

Discussion

Case 1 and her mother both had bilateral optic atrophy causing partial sightedness. Case 1 has obvious unilateral facial weakness. The mother has slight unilateral facial weakness which has been present since infancy. This pattern is compatible with autosomal dominant inheritance.

The optic atrophy in this family has an early age of onset compared with isolated dominant optic atrophy where onset is typically at 4-8 years of age and nystagmus is rare. In Hoyt’s study of 31 patients from six pedigrees with dominant optic atrophy, five had documented visual loss in infancy and four had nystagmus. He comments that some authors have considered there to be a separate genetic group with infantile onset, but in his series the four patients with nystagmus came from two pedigrees that contained a number of other affected relatives with the more classical insidious onset without nystagmus. Linkage studies have identified a locus at 3q28-qter but there is evidence of locus heterogeneity.

Non-ocular abnormalities are rare in dominant optic atrophy. Eight of the patients in Hoyt’s series had sensorineural hearing loss including four patients (from two different pedigrees) who had presented with nystagmus. There were no reports of facial palsy.

In this family, the facial palsy may be separate from the optic atrophy or it might be part of a syndrome or association. Searches of standard texts and current computerised databases did not indicate possible candidate syndromes. All syndromes listed with both optic atrophy and facial weakness are either bone dysplasias or have other neurological signs. It is likely that the early onset optic atrophy with the associated facial palsy in this family have a unifying cause. We suggest that this probably represents a new genetic entity given the absence of reports of facial palsy in families with dominant optic atrophy.

This case has been reported with the full written permission of the mother.

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