Recurrent Orbital Myositis

Report of a Familial Incidence

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Background: In orbital myositis, painful diplopia develops owing to an enlargement of the extraocular muscles. Diagnosis is established based on history, clinical manifestations, and therapeutic response to steroids, with the findings of magnetic resonance imaging providing additional information.

Observation: We observed a family in which 4 members had an ophthalmopathy suggestive of orbital myositis. The affected members are a sibling pair (female and male) and 2 children of the brothers of their father's father.

Conclusion: The familial incidence suggests a potential genetic predisposition in the development of orbital myositis.

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PERIORBITAL PAIN, swelling of the eyelid, conjunctival injection, and diplopia due to restriction of extraocular muscle movement characterize orbital myositis.1 The disorder usually begins acutely and in most cases resolves rapidly after treatment with steroids. Although it is mostly unilateral, occasionally both orbits may become involved. There are anecdotal reports of relapsing episodes of orbital myositis in an individual patient,2 but, to our knowledge, a familial occurrence of recurrent orbital myositis has not yet been described. We report on a family in which 4 members experienced episodic eye symptoms consistent with an ophthalmopathy suggestive of orbital myositis, thus indicating a potential genetic predisposition in the development of this disorder.

REPORT OF CASES

CASE 1

A 16-year-old white girl presented in April 1996 with persistent diplopia in upgaze. Orbital disease had begun 6 years earlier in January 1990 with loss of motility predominantly of the left eye, painful diplopia, eyelid swelling, tearing, and burning and redness of the left eye. The patient reported that her eyes had been prominent at that time. Her vision was perfectly normal. She was treated with prednisone, which produced an improvement of the orbitopathy over the following weeks. After the dosage of prednisone therapy was lowered, conjunctival injection, swelling of the left eyelid, and diplopia resumed. Treatment with higher doses of prednisone led to a rapid improvement of the symptoms. Although systemic steroid therapy was continued afterward at a dosage of 20 mg/d, 3 other episodes involving diplopia, proptosis, and soft tissue signs occurred mainly involving the left orbit. Notably, each of the exacerbations occurred in the winter of the respective years (January 1993, February 1994, with involvement of the right eye; and January 1995). Over the years, the patient's double vision became permanent. On examination in April 1996, she had diplopia in upgaze and lateral gaze to the left side; gaze to the right was also mildly limited. Her- tel exophthalmometry measurements were 14 mm on the right and 15 mm on the left. There was no history of thyroid disease, and, on examination, there were no clinical signs or symptoms of hyperthyroidism. The thyroid was normal in size and consistence, and the results of sonography of the thyroid were normal. The following laboratory investigations revealed no abnormalities: complete blood cell count; determination of serum urea nitrogen, electrolyte, creatine kinase, complement, C-reactive protein, immu-

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globulin fraction, and blood glucose levels; liver function tests; Westergren sedimentation rate; serum protein electrophoresis; and serologic testing for acetylcholine receptor antibody. Serum levels of free triiodothyronine, free thyroxine, thyrotropin, thyrotropin receptor antibodies, and thyroid stimulating antibody were normal, as were the results of antimicrosomal and antithyroglobulin studies and a protirelin stimulation test.

A computed tomographic scan obtained in January 1990 showed an enlargement of the left lateral rectus muscle and, to a lesser extent, the right medial rectus muscle. There was no involvement of the tendons. In January 1993, the patient was examined with magnetic resonance imaging, and enlargement of the superior rectus on the left side was demonstrated in several sections. During the next episode, in March 1994, a magnetic resonance imaging scan showed an enlargement of the lateral rectus in the right orbit (Figure 1).

CASE 2

In December 1995, the younger brother of patient 1 (Figure 2), at the age of 14 years, had a similar episode involving painful swelling of the eyelid and proptosis of the right eye 5 months before he presented for examination. He had single vision in the primary position, but had noticed diplopia on upgaze and gaze to the right. The symptoms improved gradually and completely disappeared within 3 weeks, without treatment. He had no history of thyroid disease. On examination, he had no diplopia, and there were no soft tissue signs. The results of the neurological examination were normal, and there were no clinical signs of hyperthyroidism. The thyroid was normal in size and consistence, and ultrasonography of the thyroid showed no abnormality. Results of the blood tests, including serum levels of acetylcholine receptor antibody, free triiodothyronine, free thyroxine, thyrotropin, thyrotropin receptor antibodies, and thyroid stimulating antibody were normal, as were those of antimicrosomal and antithyroglobulin studies and a protirelin stimulation test.

CASES 3 AND 4

The medical histories of 2 cousins of the father of patients 1 and 2 (children of brothers of their father’s father [Figure 2]), a 31-year-old white woman and a 26-year-old white woman, revealed similar episodes of painful diplopia. In the 31-year-old, the onset of symptoms had occurred at the age of 12 years. Since the episodes always improved rapidly after some weeks without treatment, she had never consulted a physician about the condition. After the birth of her second child, at the age of 28 years, no further episodes occurred. She had no history of thyroid disease. The results of her neurological examination were normal, and there were no clinical or laboratory signs of thyroid disease. Blood tests, which were the same as those performed in patients 1 and 2, revealed no abnormalities. The 26-year-old woman was reported by the family to have had intermittent painful diplopia. A diagnosis of myasthenia gravis had been presumed, and a thymectomy had been performed. However, no more data could be obtained regarding her history. She had moved to Australia and was not available for follow-up.

HLA typing was performed in patients 1, 2, and 3, but none of the patients had any alleles that could be an indicator of Graves disease. Figure 2 shows the pedigree of the family.

COMMENT

Orbital myositis begins acutely with unilateral periorbital pain, particularly on movement of the eye, diplopia,
and often exophthalmos. All 3 patients described herein had episodes of painful diplopia, and in all cases onset of the episodes was acute and unilateral. Systemic administration of steroids, which is the treatment of choice for orbital myositis, led to a rapid improvement of the symptoms in patient 1. However, in patients 2 and 3, the clinical manifestations disappeared after some weeks without treatment. Based on the clinical manifestations in the 3 patients, we made a diagnosis of orbital myositis. Although there is a broad list of differential diagnoses in acute diplopia, the main differential diagnosis that remained in our patients was Graves hyperthyroid ophthalmopathy. In Graves ophthalmopathy, there is usually some indication of thyroid dysfunction. However, in about 10% of cases, no clinical or laboratory evidence of thyroid disease can be identified. The patients in this subgroup were first described in 1941, and the disorder was termed euthyroid ophthalmopathy. None of our patients had a history of thyroid disease or any clinical or laboratory evidence of thyroid dysfunction on careful examination. HLA typing showed no alleles that have been suggested to be associated with Graves disease. In euthyroid ophthalmopathy, however, the ocular changes may follow an independent course from the development of thyroid dysfunction, and they may occasionally precede evidence of such abnormalities by months or years. Since the follow-up of our patients has lasted several years, it is increasingly improbable but cannot be excluded that they may develop thyrotoxicosis in the future.

Magnetic resonance imaging may, apart from excluding other potential differential diagnoses such as tumor infiltration of the orbit, provide additional diagnostic information. In patient 1, it showed the characteristic finding of enlargement of a single extraocular muscle. It has been repeatedly suggested that magnetic resonance imaging may help to distinguish between orbital myositis and endocrine ophthalmopathy. It is believed that in euthyroid ophthalmopathy the swelling of the extraocular muscles is confined to the muscle belly, which shows a smooth contour, while the tendon (between the anterior muscle belly and the globe) remains spared. In contrast, in orbital myositis, the affected muscle shows an irregular contour and the inflammation may extend to the orbital fat. There is usually an indication of involvement of the tendon. Although in the index patient, no involvement of the tendon was found on computed tomographic or magnetic resonance imaging scans, the history, clinical findings, and therapeutic response established the diagnosis of orbital myositis. The absence of the “tendon sign” does not exclude the diagnosis of orbital myositis.

Relapsing episodes of orbital disease have been documented in cases of orbital myositis. Several episodes showed a seasonal occurrence, as was observed in one of our cases (patient 1), and followed nonspecific upper respiratory infections. All exacerbations occurred in winter in patient 1, although a preceding upper respiratory infection had not been noted at that time.

To assess the likelihood that multiple affected family members have orbital myositis or Graves disease by chance rather than by descent, it would be useful to state the population indices of orbital myositis and of Graves disease. However, to our knowledge, there are no data available on the population frequency of orbital myositis. Since only 3 affected members of the family were examined and no thorough investigation of the large family was feasible, a reliable pedigree analysis cannot be provided. It is known that ophthalmopathy occurs in only 5% of patients with Graves disease, and of this subgroup of patients only about 10% have no demonstrable thyroid disease.

In conclusion, in our patients, the clinical presentation and ancillary findings favor the diagnosis of recurrent orbital myositis. To our knowledge, this is the first report of a familial incidence of this disorder. Unless more families turn up with similar symptoms, however, one cannot be sure that this is genetic transmission. Nevertheless, it seems appropriate to consider that a genetic predisposition may represent a relevant factor in the development of orbital myositis. Other events may influence the expression of the disease.

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