FUCHS' HETEROCHROMIC CYCLITIS AND RETINAL VASCULAR ABNORMALITIES IN PROGRESSIVE HEMIFACIAL ATROPHY

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SUMMARY

We report a case of progressive hemifacial atrophy with a combination of ipsilateral Fuchs' heterochromic cyclitis and retinal vascular abnormalities. Recently, evidence was found for a neurovascular defect in hemifacial atrophy. Our case not only supports the (clinical) association between Fuchs' heterochromic cyclitis and hemifacial atrophy, but the retinal vascular abnormalities found in this patient add further support to the existence of a neurovascular defect. These findings and our short review of the literature point to the hypothesis of a common sympathetic defect, implicated in the aetiology of both Fuchs' heterochromic cyclitis and progressive hemifacial atrophy. It has to be borne in mind, however, that Fuchs' heterochromic cyclitis has been reported in association with other diseases. It seems likely that although Fuchs' heterochromic cyclitis is a single clinical entity, it may have more than one cause.

Hemifacial atrophy (Parry-Romberg syndrome) was first described by Parry in 1825.¹ A more detailed description of this syndrome was made by Romberg in 1846, who distinguished it from the congenital non-progressive form of this disorder.² Hemifacial atrophy is a rare condition of unknown aetiology, characterised by a slowly progressive atrophy of one side of the face, primarily involving skin, subcutaneous fat and muscles. The onset of the disease usually occurs in the first two decades of life. If the onset is before the age of 10 years, the bone is also involved, resulting in a severe facial deformity.³ In 7% of cases the ipsilateral limbs and trunk are involved and occasionally the syndrome is bilateral.⁴⁻⁶ Females are more often affected than males in a ratio of 3:2.⁴ Progression of the disease occurs more rapidly in the first 2-10 years after onset and it may stabilise at any age.⁷

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Many hypotheses have been put forward to explain the aetiology of progressive hemifacial atrophy, which is still unknown. A relationship with either localised scleroderma, trauma, infection, an autoimmune disorder, a trigeminal neurovasculitis, hereditary degenerative factors or a defect of the sympathetic nervous system have been suggested as possible causes.^{3,5,6,8,9} Ocular involvement in hemifacial atrophy occurs in up to 40% of cases.¹⁰ Most of the ophthalmologic conditions appearing in hemifacial atrophy have been described in single case reports.

We describe a patient with progressive hemifacial atrophy who developed the clinical features typical of Fuchs' heterochromic cyclitis. In addition, ipsilateral retinal vascular abnormalities were present in the fundus of this same patient. This combination has not been reported before in the literature. Our case not only supports the association between Fuchs' heterochromic cyclitis and hemifacial atrophy, but the retinal vascular abnormalities found in this patient add further support to recent evidence of a neurovascular defect in progressive hemifacial atrophy.

CASE REPORT

A 28-year-old Caucasian woman with a previous diagnosis of right-sided progressive hemifacial atrophy was referred to us several years ago because of visual complaints in the right eye. The process of facial atrophy had started when she was 19 years old. Because of this late onset there was no bone involvement and only slight cartilage involvement. The skin, subcutaneous fat and muscles, however, were severely affected and an obvious facial asymmetry was visible (Fig. 1). On examination an enophthalmos and thinning of the eyebrow were noted on the right side. Best visual acuity was 10/20 for the right eye and 20/20 for the left eye. The pupil of the right eye was larger and it showed a delayed pupillary reaction to light. In the right eye small translucent keratic precipitates scattered over the entire endothelium and minimal aqueous flare and cells were seen. Diffuse iris atrophy and heterochromia were noted. No posterior synechiae were

Fig. 1. Conspicuous facial asymmetry in a patient with right-sided progressive hemifacial atrophy, Fuchs' heterochromic cyclitis and retinal vascular abnormalities of the right eye.

present. The lens was clear and in the anterior vitreous fine dust-like opacities were present. On the basis of these characteristic findings the diagnosis of Fuch's heterochromic cyclitis was made. On fundus examination a haemorrhage above the macula along the superotemporal artery and exudates in the upper nasal quadrant of the macula were seen (Fig. 2). Fluorescein angiographic studies revealed the presence of macroaneurysms of the superotemporal artery with leakage in the direction of the macula (Fig. 3). Laser coagulation was performed to decrease this leakage. In the 3 years that followed the keratic precipitates and minimal aqueous chamber reaction remained unaltered. The haemorrhages and exudates diminished after repeated laser coagulation.

Fig. 2. Fundus photograph of the right eye showing haemorrhage (arrow) and macroaneurysms (arrowheads) along the superotemporal artery, and exudates in the upper nasal quadrant of the macula.

DISCUSSION

The combination of Fuchs' heterochromic cyclitis and retinal vascular abnormalities as found in our patient has, to our knowledge, not been reported before in hemifacial atrophy. Recently Gass *et al.*¹¹ were the first to describe four cases of hemifacial atrophy with similar retinal vascular abnormalities. In one of their patients a mild cellular reaction in the anterior chamber and vitreous was noted. A second patient had segmental atrophy of the iris. No other anterior segment abnormalities were described. In our patient, however, in addition to the retinal vascular defects, anterior segment findings typical of Fuchs' heterochromic cyclitis were found: small translucent keratic precipitates scattered over the entire endothelium, minimal aqueous flare and cells, diffuse iris atrophy, discrete heterochromia, fine opacities in the anterior vitreous and no posterior synechiae. Complicated cataract or glaucoma had not (yet) developed.

Fuchs' heterochromic cyclitis was first considered to be associated with hemifacial atrophy because both diseases were said to be part of the status dysraphicus.¹² Sugar and Banks¹³ reviewed the 13 cases of Fuchs' heterochromic cyclitis in hemifacial atrophy reported since 1913 and added another case from their own practice. They suggested that both diseases may result from neurovascular or neurotrophic changes, caused by disturbances of the sympathetic nervous system. Loewenfeld and Thompson¹⁴ objected that the majority of descriptions of Fuchs' heterochromic cyclitis found in cases with hemifacial atrophy were not typical: there were unusual (pigmented) corneal precipitates, there was no iris atrophy or heterochromia, and in some cases posterior synechiae were found. In our patient, however, the typical features of Fuchs' heterochromic cyclitis were present, as mentioned above. Despite the arguments put forward by Loewenfeld and Thompson¹⁴ in their extensive review to reject the association between Fuchs' heterochromic cyclitis and hemifacial atrophy, and the hypothesis of a sympathetic

Fig. 3. Fluorescein angiogram of the right eye showing macroaneurysms (arrows) of the superotemporal artery.

defect implicated in the aetiology of both diseases, many authors¹⁵⁻²¹ still support this theory put forward by Sugar and Banks.¹³

Moss and Cirkelair¹⁵ provided evidence for this sympathetic hypothesis by demonstrating hemifacial atrophy after unilateral sympathectomy in young rats. Pupillary changes, Horner's syndrome and heterochromia are often reported in patients with hemifacial atrophy, and are all signs of an impaired sympathetic nervous system.¹⁶

In 1953 a case was described in which Horner's syndrome and Fuchs' heterochromic cyclitis developed consecutively in the same eye after a stellate ganglionectomy.¹⁷ Recently, five patients with unilateral Fuchs' heterochromic cyclitis and ipsilateral Horner's syndrome were reported.¹⁸ Electron microscopic studies on Fuchs' heterochromic cyclitis have suggested that the heterochromia may result from a defective melanin production due to abnormal adrenergic innervation.¹⁹ Furthermore, it has been postulated that the vascular leakage seen in the iris fluorescein angiographic studies on Fuchs' heterochromic cyclitis may be caused by a disturbance in the iris vessel innervation, which is derived solely from the sympathetic nervous system.^{20,21}

In a recent electron microscopic study evidence was found for a neurovascular defect involved in the pathogenesis of hemifacial atrophy.³ The retinal vascular abnormalities seen in our patient and in those of Gass *et al.*¹¹ may result from such a neurovascular defect. These findings and the above mentioned studies point to the hypothesis of a common sympathetic defect implicated in the aetiology of hemifacial atrophy and Fuchs' heterochromic cyclitis.

In addition to hemifacial atrophy, Fuchs' heterochromic cyclitis has been reported in association with other conditions: ocular toxoplasmosis,^{22,23} ocular trauma,²⁴ retinitis pigmentosa²⁵ and the subclavian steal syndrome.²⁶ Moreover, recent immunohistochemical studies on iris biopsy specimens^{27,28} have failed to show any specific abnormalities in these patients. It is therefore conceivable that although Fuchs' heterochromic cyclitis is a single clinical entity, it may have more than one cause.

Key words: Fuchs' heterochromic cyclitis, Hemifacial atrophy, Retinal vascular abnormalities.

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