

A presumed case of functional visual loss with eventual diagnosis of Stargardt's disease

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Abstract

Aim: To discuss a presumed case of functional bilateral amblyopia which had an eventual diagnosis of Stargardt's disease.

Methods: Details are reported of a girl who presented to the Orthoptic Department at age 9 years having previously attended the orthoptic department as a young child. Documentation of the case is presented including the findings in childhood, the findings at later presentation and the findings when an eventual diagnosis was made.

Results: The orthoptic findings for this girl when discharged at age 8 years differed considerably from the findings at age 9. When discharged at age 8 years she showed acuities of 6/6 right and 6/12 left. A minimal residual left esotropia was present. At re-presentation 7 months later visual acuities had dropped to 6/24 right and left. No cause for the reduction in acuity could be found.

Conclusions: Stargardt's disease is an inherited condition which affects the macula. There is usually considerable sight loss although complete loss of vision is rare. Early in the disease the macula and the electroretinogram can appear normal, which slows the initial diagnosis. Subsequently children may be suspected of malingering. In time the characteristic changes in the retina facilitate a diagnosis.

Key words: Electrodiagnostic tests, Functional visual loss, Stargardt's disease

Introduction

Stargardt's disease (juvenile macular dystrophy) is an inherited condition. Inheritance may be autosomal dominant, autosomal recessive or X-linked.¹ It is characterised by visual loss and macular atrophy. Presentation is in the first to second decade of life with gradual and progressive visual loss.² The loss of acuity may present before or be out of line with macular changes, and therefore patients may be suspected of malingering.³ Once the visual acuity reduces below 6/12, there is usually a rapid decrease to about 6/60, where most cases stabilise. Retinal signs are progressive. Initially the fovea may be normal or show non-specific

mottling. A foveal appearance described as 'beaten bronze' later develops, followed by geographic atrophy which may have bull's-eye configuration. Fluorescein angiography shows an absence of normal background fluorescence. If geographic atrophy is present, a window effect at the macula is shown. The condition is usually stable by early adulthood.

Case report

A 9-year-old girl presented to the orthoptic department following a referral from her optician. She had attended her optician for a routine sight test and had complained that her acuity had decreased recently. The optician reported that corrected acuities had been 6/6 right and 6/12 left in September 1999 but had reduced to 6/24 either eye in July 2000. There was no significant change in prescription and ophthalmological examination was normal. He concluded that the reduction in vision was due to stress and reassured the patient that things would improve. This did not happen on subsequent testing and therefore the child was referred back to the hospital eye service for further investigation.

The girl had first attended the orthoptic department when she was 18 months old and the parents had noted a left esotropia. There was no information on family history as the child was adopted. Cycloplegic refraction revealed mild hypermetropia which was fully corrected: right +1.50DS, left +2.00DS. She was seen regularly in the orthoptic department until she was 8 years old. Occlusion therapy was carried out on an ongoing basis and discontinued at 8 years old. At 3 years old a left medial rectus recession and left lateral rectus resection were carried out. At discharge in 1999 corrected acuities were right 6/6 and 6/12 left. A minimal left esotropia was present with no demonstrable binocular functions. Review by an ophthalmologist had been performed on an annual basis until discharge and no abnormality of fundi were ever noted.

When the girl re-presented to the Orthoptic Department aged 9 years full orthoptic and ophthalmic investigations were carried out. Orthoptic status had not altered but acuities had deteriorated to 6/24 right and left. It was noted that the child's parents had recently separated and that her new stepfather had recently been diagnosed with complicated keratoconus. Therefore there was a considerable amount of stress within the family. Since no reason could be found for the visual loss a tentative diagnosis of functional visual loss was made. The patient was referred to another centre for electrodiagnostic tests, including occipital pattern visual

evoked potentials (VEPs) and pattern and flash electroretinograms (ERGs), to ensure nothing had been missed. Visual fields and colour vision were analysed. Both are recorded as being difficult to test, with the results being somewhat inconclusive. Electrodiagnostic tests were returned as normal. The patient continued to be reassured that her acuity would improve and was discharged to be seen when necessary.

Between 2001 and 2006 she attended approximately annually. She continued to complain of reduced vision and was concerned regarding the worsening appearance of her consecutive divergent squint. The patient's mild hypermetropic glasses were discontinued as they did not improve her vision and made the divergent squint marginally worse. Further surgery was not advocated as the angle of deviation was not considered large enough.

Numerous appointments were made by the child's parents with different opticians to try to assist in a diagnosis between 2001 and 2006. No further information was ever gained.

In view of the persistent complaint of reduced acuity in the absence of any pathology a tentative diagnosis of functional visual loss was made. The girl was referred to the paediatric and mental health services. The paediatric team stated in 2006 that there was a remote chance that this could be a case of temporal lobe epilepsy. Consultation with the mental health team in 2004 concluded that the girl's parents should try to reduce pressure on her but at the same time ensure that she was not overwhelmed with attention. The message was reinforced that the child had perfectly adequate functioning vision and she was encouraged to take more responsibility for her actions.

In 2007 the child's mother decided to visit another optician to try to achieve a diagnosis for the visual loss. This visit revealed fundal changes which led to a diagnosis of Stargardt's disease. Referral for electrodiagnostic tests confirmed this diagnosis to be correct.

Conclusion

The presentation of Stargardt's disease is often such that the visual acuity deteriorates a long time before fundal abnormalities are detected, thus leading to a diagnosis of suspected malingering.

Orthoptists frequently deal with cases of functional visual loss and deal with them appropriately. A diagnosis of Stargardt's disease should be suspected in cases of

prolonged, undiagnosed, unresolving visual loss. The eventual diagnosis of Stargardt's disease was made even more difficult in this particular case due to the lack of family history information since the child was adopted. Since diagnosis it has come to light that the patient has a natural brother with Stargardt's disease.

In this particular case the delay in reaching a diagnosis meant that referral to the visual impairment team was not made, access to low visual aids was not given, and educational assistance such as extra time for examinations was denied.

There was a 7-year delay between re-presentation and diagnosis, the consequence being that a lot of essential schooling was missed and a 'malingering' label was wrongly applied.

Comment

- Orthoptists are expert at examining patients who are suspected of having functional visual loss. In a majority of cases a cause for the 'visual loss' is established, the patient is reassured and over time normal acuity recordings are made.
- Be vigilant about cases of suspected malingering that fail to recover over time.
- Be aware that the retinal and electrodiagnostic signs in Stargardt's disease often become manifest a long time after visual loss begins to occur.
- Document family histories of blindness or visual loss carefully, as this may give an indicator to the eventual diagnosis.
- Perhaps it is pertinent to consider repeating electrodiagnostic tests after 2–3 years in the absence of a definitive diagnosis for persistent visual loss.
- Optical coherence tomography (OCT) may be performed, where available, to visualise the layers of the retina and assist in a diagnosis.

Sincere thanks are due to Mr Hugh Bacon, Consultant Ophthalmologist, and the patient for permission to present this case.

References

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