Behavioural disturbance as a manifestation of ocular disease in children

EDITOR—Ocular disease in young children may present with behavioural changes and psychological disturbance. We highlight two exquisitely accurate cases of Leber's hereditary optic neuropathy, and discuss the difficulties faced in the diagnosis and management of these cases, and to emphasise the importance of meticulous clinical examination and appropriate investigation to exclude any organic visual disorder before a case is erroneously labelled 'functional' in origin.1

CASE REPORTS

Case 1

A 5-year-old boy presented with a history of increasing clumsiness. His parents also commented on worsening behaviour with frequent temper tantrums and episodes of intense rage. Family history and neonatal history were negative. Visual acuity was 6/24 right, 6/36 left and there were no abnormalities detected in either eye. Electrodiagnostic tests were normal and it was thought that the problem was probably functional in nature. He was transferred to the Children's Hospital in Birmingham for further assessment when it was found that his vision had decreased to 4/60 in each eye and both optical discs were pale. Aryl sulphatase levels were normal as was magnetic resonance imaging scan, ruling out metachromatic leukodystrophy. Visual evoked responses (VERs) were now reduced and pattern VERs showed marked delays in both eyes. Electroretinograms (ERGs) were normal. These findings were consistent with an optic neuropathy and mitochondrial DNA analysis revealed a mutation at nucleotide position 3460, confirming the diagnosis of Leber's hereditary optic neuropathy (LHON).1,3

Case 2

A 7-year-old boy was seen at the Birmingham and Midland Eye Hospital with difficulties focusing near vision. He was aggressive, badly behaved, and was threatened with expulsion from school. He had initially been seen elsewhere at 4 years of age with a left convergent squint, bilateral hypermetropia, and astigmatism in the left. Glasses and occlusion therapy were given and best corrected vision at that time was 6/6 right and 6/18 left. An abnormal macula reflex was also noted but it was felt that the squint and anisometropia were sufficient to explain his vision. When seen 3 years later, his best corrected vision had deteriorated to 6/36 right and 6/12 left but there was no manifest squint with glasses. Near vision was N8 bilaterally. Macular abnormalities were noted in both eyes consisting of a pale yellow lesion in the left and some localised pigmented disturbance in the right. Electro-oculogram (EOG) was grossly abnormal at 102% right and 100% left. ERGs were normal. This was consistent with Best's vitelliform dystrophy.4,5

COMMENT

The cases reported are two examples of young children with behavioural disturbance and reduced visual acuity. In each case, the child was otherwise healthy, there was neither intellectual deterioration nor any obvious ocular pathology on examination. Their family histories did not raise any suspicion of a visual handicap. This clinical scenario is usually encountered in preschool or school age children and is a diagnostic challenge. There is the temptation to attribute the visual defect to emotional and psychological factors, dismissing the problem as non-organic in origin. There is little literature on cases like these but we believe that they may be more common than is suggested by the lack of reports. A further two cases with X linked retinoschisis who presented similarly were seen in Birmingham over the past year (Fielder AR, personal communication). Our report draws attention to the fact that it is essential that these cases are examined and investigated in order to identify any underlying pathology.

Our first patient is the youngest case of LHON reported. This is a maternally inherited disease caused by a specific mitochondrial DNA mutation that presents typically in adolescent and young adult males with central visual loss.2 Our case presented atypically as the child was too young to complain of visual loss and his visual ability and subsequent frustration manifested in temper tantrums and rages. The only abnormal finding was pallor of both optic discs. Electrodiagnostic testing was informative in initial examination and the diagnosis was established using mitochondrial DNA analysis. In our second case, the EOG and ERG were indispensable in confirming the diagnosis of Best's vitelliform dystrophy. Several clinical presentations have been described,4 but unless the patient is examined during the vitelliform phase where the lesion is characteristic, the diagnosis may be missed. Both cases underscore the importance of electrodiagnostic testing in children as an excellent objective test that is independent of the patient's cooperation, intelligence, and literacy. Mitochondrial DNA analysis was performed to make a definitive diagnosis of LHON in our second case. Mitochondrial DNA has implications for visual prognosis and counselling.2,5 Aryl sulphatase levels were also checked to exclude metachromatic leukodystrophy which presents in young boys with behavioural changes, intellectual deterioration, and optic atrophy. Visual disorders in children can present in different, sometimes obscure ways. We should always be aware that behavioural disturbance may be one. The absence of a family history or typical clinical features can at times be misleading. It is imperative that the child undergoes adequate ophthalmic investigation, and the responsibility lies with the clinician to differentiate between organic and non-organic disease.1

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Misleading aspiration biopsy in combined intraocular pathology

EDITOR—Occasional misinterpretations of aspiration biopsy in presumed intraocular malignancies are mainly due to sampling errors.1–3 The presence of different palpatological processes within the same nodule is a pitfall of the sampling error that has not been emphasised in the literature but is reported here.

CASE REPORT

A 59-year-old woman presented to her local ophthalmologist with a 3 day history of deteriorating vision and a 2 week history of flashing lights in her left eye. The visual acuity was hand movements and the intraocular pressure was 13 mm Hg. On examination a large inferior exudative detachment, that did not transilluminate nasally, was noted. The patient was referred to the Tennent Institute of Ophthalmology 9 days later. On ultrasound an inferior mass 7-8 mm in thickness with a basal diameter of 16-5 mm was identified. The reflectivity of the tumour was not typical of a choroidal melanoma and a possible metastatic origin was suggested. The systemic examination was negative, and a diagnosis of a presumed choroidal melanoma was retained. Thus, the patient was taken to the operating theatre for an attempt at intraoperative transillumination the examination of the choroid after the preparation of the scleral flap showed no evidence of a tumour. Subretinal fluid was aspirated for histology and the sclera was closed.

Two smears and cytospin preparations revealed the presence of lysed and intact red blood cells, haemangiomacrophages (Fig 1a), a few inflammatory cells, and a population of histiocytoid cells containing neuro-melanosomes (Fig 1b). The latter cells were felt to be retinal pigment epithelial (RPE) cells or RPE derived macrophages. Thus, an organising haemorrhage arising in a disciform degeneration was indicated in the report.

A vitreous haemorrhage developed. A magnetic resonance image was inconclusive, but a vitreotomy revealed an extensive central disciform lesion (Fig 1c), a similar lesion in the periphery (Fig 1d), and a large inferotemporal exudative retinal detachment. A fluorescein angiogram confirmed these findings. The vitreous aspirate obtained at surgery contained red blood cells only.

The patient ignored her follow up appointment but returned 4 months later with an enlarging supranasal tumour. The ultrasound was now diagnostic of choroidal...