SUDDEN DEVELOPMENT OF BILATERAL CATARACT IN A CHILD WITH DUBOWITZ SYNDROME: A CASE REPORT

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SUMMARY

The Dubowitz syndrome is a rare, autosomal recessive disorder including intrauterine growth retardation, craniofacial abnormalities, mental retardation and eczematous skin eruption. Ocular problems occur in about 20%: strabismus, blefarophimosis, ptosis, telecanthus and epicanthal folds being the most frequent ones. We present a three years old girl, diagnosed with Dubowitz syndrome, with sudden visual loss due to bilateral cataract.

RÉSUMÉ

Le syndrome de Dubowitz est une affection rare à transmission autosomale récessive. Il se caractérise par une croissance intrautérine retardée, des anomalies craniofaciales, une retardation mentale et une éruption eczémateuse de la peau.
Des problèmes oculaires se manifestent dans 20% des cas. Parmis eux le strabisme, le bléfarophimosis, le ptosis, le télécanthus et les plis épicanthaux sont les plus fréquents.
Nous présentons le cas d'une fillette de 3 ans, avec syndrome de Dubowitz, qui se présente avec une perte visuelle aiguë due à une cataracte bilatérale.

KEY WORDS

Dubowitz syndrome, cataract

MOTS CLÉS

Syndrome de Dubowitz, cataracte

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INTRODUCTION

A three years old girl, diagnosed with Dubowitz syndrome, presented at our department with sudden visual loss due to bilateral lensopacification. The Dubowitz syndrome is a rare, autosomal recessive disorder including intrauterine growth retardation, craniofacial abnormalities, mental retardation and eczematous skin eruption (3,6). It seems to have a wide range of expressions (5). The most frequently encountered craniofacial features are microcephaly, sparse hair, a high forehead, eye abnormalities, broad nose, palate anomalies, micrognathia/retrognathia and dysplastic ears (2). Mild to moderate mental retardation is observed in about 50% of patients. Abnormal behaviour is rather common, and hyperactivity has been described in 40% of patients (1).
Ocular problems occur in about 20%: strabismus, blepharophimosis, ptosis, telecanthus and epicanthal folds being the most frequent ones (3).
Our patient is the second reported child with Dubowitz syndrome and bilateral cataract. The first patient has recently been described by Rodden et al (3).

CASE REPORT

The girl was born at term to a 34 years old gravida-2, para-1, abortion-0 mother after an uneventful pregnancy and delivery. The birth weight was 2,1 kg (<3th percentile). She measured 44 cm (3<th percentile) and the head circumference was 30 cm (3<th percentile). Clinical examination showed a peculiar facial appearance with a ptotic right eye, clinodactyly of one finger and a sacral dimple (fig 1). At the age of 10 months the microcephaly became more obvious. There was poor weight gain due to severe feeding problems. She also had severe eczema, a high forehead, a wide nasal bridge and sparse thin hair. She was developmentally retarded and had severe behavioural problems. The patient's clinical findings were consistent with the Dubowitz syndrome. Ophthalmological examination at the age of 18 months showed a right ptosis with no occlusion of the pupil. Biomicroscopic examination of the anterior segment and fundoscopy were normal. Cycloplegic retinoscopy revealed + 3 sphere at the right eye and + 3,5 sph + 2,5 cyl/90° at the left eye. Occlusion therapy of the left eye was initiated to improve vision in the ptotic right eye. At the age of three years biomicroscopic examination of the anterior segment revealed discrete bilateral subcapsular opacities, not interfering with vision. Fundoscopic examination was normal. Two months later, she suddenly lost vision in both eyes. She could't find her way at home and only reacted to sound. Ophthalmological examination revealed bilateral total cataract (fig 2). Lensaspiration with posterior rhexis and lensimplantation was performed in both eyes 3 months apart. Preoperative readings revealed axial lengths of 20.9 mm and 20,2 mm. Keratometry readings showed a mean value of 46,5 diopters at the right eye and 48 diopters at the left eye. Lensimplantation was performed with a + 26 D lens at the right eye and a + 27 D at the left eye. After the operation the visual functions recovered. Visual acuity testing with Fooks test was not possible. She could recognize objects of 0,5 mm diameter at 30 cm distance with the left

Fig. 1: In this patient with Dubowitz syndrome ptosis of the right eye is evident.

Fig. 2: Bilateral cataract in a patient with Dubowitz syndrome.

eye. The right eye is amblyopic. Three months later ptosis correction of the right eye was performed.

DISCUSSION

The Dubowitz syndrome was first described by Dubowitz in 1965 (2). Its pathogenesis is unknown. It is presumed to represent the homozygous state of an autosomal recessive mutation. The phenotypic variability appears to be very broad, suggesting action of many modifying genetic or epigenetic factors (4). The clinical findings in our patient were consistent with Dubowitz syndrome: small stature, microcephaly, ptosis, wide nasal bridge, sparse thin hair, eczema, mental retardation, hyperactivity, poor feeding and skeletal abnormalities.

The ophthalmological findings of telecanthus, strabismus, epicanthal folds, ptosis and blepharophimosis have been well documented in Dubowitz syndrome (3). Other ocular abnormalities such as microphthalmos and coloboma have also been described (4).

We report the second patient with Dubowitz syndrome associated with bilateral cataract. Visual loss developed over a few days and at very young age. The patient described by Rodden et al (3) developed slowly progressive cataract at the age of 12 years. The etiology of the lensopacification is not known but the surface ectodermal derivation of the lens may point to the possibility of a gene related to connective tissue formation or regulation (3).

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