Delayed Visual Maturation – A Clinical Case

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SUMMARY

A first eye examination is performed on a two and a half month old patient who presents with no visual response to fixation or to pursuing a target or a light despite an unremarkable ocular health assessment. This temporary eye condition is known as Delayed Visual Maturation (DVM).

CONCLUSION: A subsequent eye examination was performed at age five and a half months only to reveal that the child's visual behaviour was now considered to be appropriate for his age. The diagnosis of DVM remains a diagnosis of exclusion and requires a comprehensive eye examination, electrodiagnostic testing and imaging techniques which may eventually include MRI.

Introduction

Suspected abnormal vision in a newborn should not be taken too seriously. In fact, in 80% of cases, the professional discovers, upon examination, an obvious cause of visual impairment such as a cataract, glaucoma or albinism.[1] Sometimes, as with Leber congenital amaurosis, it is instead marks on the retina, pupil anomalies or nystagmus. Other anomalies, such as cortical blindness, are accompanied by major neurological disorders. However, if it is simply delayed visual maturation, all the objective tests may be normal, which complicates the diagnosis. Electrodiagnostic testing then becomes very helpful for making a differential diagnosis between the various conditions and guiding the parents regarding the permanent or temporary aspect of the reduced vision.

Case report

A.J., a boy at two-and-a-half months of age, is brought in by his mother for a comprehensive eye examination. She noticed that A.J. seems to have a vision problem because he does not react to visual stimuli. He smiles when he hears human voices, but does not react to faces. A.J.'s history is otherwise unremarkable: term pregnancy with no complications, good health, no family history of systemic or eye problems. Upon examination, visual acuity via preferential looking, the ability to look at and track a light or large object, the presence or absence of nystagmus, pupillary reflexes, external examination, the ocular fundus by direct and indirect ophthalmoscopy and retinoscopy under cycloplegia are assessed (*table 1*). No particular anomalies were detected.

Given the young age of the patient and the absence of any visible ocular pathology, a follow-up in one-and-a-half months was then recommended. At that follow-up, the same results were obtained upon examination.

What is the most likely diagnosis?

Discussion

An infant with delayed visual maturation (DVM) does not fix or follow a target or light, despite an eye exam that may be completely normal. In most cases, the infant does not blink at the approach of a bright light or object. Normally, the fixation reflex may be seen in the infant at 32 weeks gestation.[2,3] At that age, preferential looking is also present.[4] In fact, newborns will prefer to look at a pattern or moving target rather than a uniform surface. Horizontal

TABLE 1: Tests and Results of A.J.'s Eye Exam

Tests	Results
Visual acuity (Teller card)	Nil
Fixation	Absent
Motility	Nil
Nystagmus	Absent
Pupillary reflexes	Normal
External exam	Normal
Ocular fundus	NO, normal retina and macula
Refraction (retinoscopy under cycloplegia)	OD +1.25/-0.50 x 180 OS +1.50

TABLE 2: Types DVM[9]

Types	Characteristics
1	Isolated DVM
2	DVM and mental retardation or epilepsy
3	DVM and primary visual anomalies

TABLE 3: Types of DVM[10]

Types	Characteristics		
1	DVM is the primary anomaly	1a	Normal perinatal period
		1b	Abnormal perinatal period
2	DVM and lasting neurological development disorder		
3	DVM, nystagmus and albinism		
4	DVM and bilateral congenital eye disorders		

tracking is present at birth, but vertical tracking develops between four and six weeks of age. Patients with DVM are typically between two and four months old and are in good health. They generally start to fix and follow objects at around six months of age. DVM is not a new condition. As early as 1926, Beauvieux was describing infants who showed little or no reaction to visual stimuli at birth.[5] Since then, a number of authors have described similar cases using different terms: *myelogenosis retardata* , *disscociated visual development* or *delayed visual development*.[6,7] Illingworth (1961) was the first to introduce the term delayed visual maturation. He described the condition of two infants, with no developmental delay, whose normal reaction to visual stimuli did not appear until six months of age.[8] En 1981, Uemera et al. introduced a DVM classification having three categories (*table 2*).[9] Fielder et al. altered that classification by instead suggesting four types with subcategories (*table 3*).[10]

DVM is an exclusion diagnosis requiring a comprehensive eye exam, electrodiagnostic testing and potentially magnetic resonance imaging (MRI). The use of visual evoked potentials (VEP) is common. However, the results vary considerably from one study to the next. Some studies have reported that infants with DVM also show an initial VEP deficit. *Flash* VEPs may be absent[7], show latency[11] or an abnormal shape[10] in the first months of life with gradual improvement after that. *Pattern* VEPs show similarities, but vary greatly depending on the stimulus used. The electroretinogram (ERG) is abnormal only if the DVM is associated with retinal anomalies.[12] The main differential diagnoses are shown in table 4. All these conditions have a comparable initial clinical presentation. The two conditions that are the hardest to rule out are bilateral pupillary hypoplasia and cortical blindness. In the first instance, the symmetrical aspect of the optic nerves can result in the pathology going undetected; the diagnosis of the second condition, which is not always easy to rule out at a young age, often requires an MRI.

The prognosis of infants with delayed visual maturation varies considerably from one study to the next and greatly depends on the type of pathology. Several authors report isolated cases of DVM[7, 11, 13] while others find them associated with other neurological disorders.[1] The isolated cases end up correcting themselves on average around the 14th week according to Fielder[10] and between three and eight months according to most of the authors.[12, 14, 15] That recovery is fast. In a few days, the infant wakes, is interested in and smiles at its surroundings. The comprehensive eye exam is then comparable to that of other normal infants of the same age. When DVM is associated with mental retardation (type 2 according to Fielder), the prognosis is not as good because of the associated neurological anomalies. When DVM is associated with another ocular defect such as albinism, aniridia or something else (type 3 and 4 according to Fielder), initial vision seems totally absent or less than what the detected ocular defect suggests. However, in a few months, on average five months, some vision appears, corresponding with what would be expected with the associated anomaly.[10]

A number of hypotheses were put forth in the past for determining the cause of DVM. According to Beauvieux, it was caused by myelination delay.[5] It was long considered that the rear visual pathways were completely myelinated in the first months of life.[16] More recent studies show that the fibres of the optic nerve start myelination at birth, but

TABLE 4: Main Differential Diagnoses

Conditions	Characteristics
Leber congenital amaurosis	Frequent nystagmus Pupillary anomaly Retinal variation Major ERG alteration Total or severe VEP deficit
Albinism	Possible nystagmus Ocular fundus hypopigmentation Normal ERG and VEP Iris transillumination enables diagnosis
Cone dystrophy	Major ERG alteration VEP affected Early nystagmus and photophobia
Bilateral pupillary hypoplasia	Normal ERG VEP +/- affected **CAT scan enables diagnosis**
Cortical blindness	Normal ERG Major VEP deficit **MRI enables diagnosis**

that the process may continue until two years of age.[17]

According to Hoyt, however, DVM was caused by a delay in the maturation of certain cortical functions[18], whereas Tresidder believes it is an abnormality in the extrageniculate system predominating in the first two months of life, while the purely cortical functions emerge after that time.[19]

DVM is a relatively rare visual condition where the infant does not react to visual stimuli, but shows no visible anomalies. DVM can be seen isolated or accompanied by other abnormalities. In most cases, visual behaviour becomes normal around six months of age. After that, or if the parents are concerned, electrodiagnostic testing (VEP and ERG) is recommended to rule out any other visual impairment.

Conclusion

At five-and-a-half months of age, A.J. was examined again. He presented with visual behaviour within the normal limits for an infant of his age. The mother noticed that he reacts normally to visual stimuli.

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