5-1-2003

Ocular motor apraxia: A familial case report

Tawna Roberts
Pacific University
Ocular motor apraxia: A familial case report

Abstract
BACKGROUND: Ocular Motor Apraxia (OMA) is a congenital ocular-motor anomaly involving the intermittent inability to initiate horizontal saccades. Smooth pursuit movements are usually normal in these patients. Signs of OMA are most apparent during infancy. Compensatory behaviors include head thrusting, blinking, and tilted head posture. Previous studies have shown that most affected children are slow in attaining early developmental milestones, and later tend to be clumsy. Difficulties in speech were very common and reading difficulties are also very well recognized in this condition. OMA is considered rare but there are no prevalence studies. The literature has previously suggested that OMA is not a genetic anomaly.

CASE REPORT: Eight month old, MR, was brought in for a vision exam because her mother was worried of possible visual dysfunction. Only positive health history is bilateral sensory-neural hearing loss (BSNHL). Visual acuity was 20/1100 OU with Preferential Looking Cards. Retinoscopy revealed low hyperopia OU and visual fields full OU. Fixation was equal and central OD and OS. Vertical saccades and pursuits are normal. Horizontal pursuits were normal following abnormal head thrusting due to lack of horizontal saccades. Anterior segment and posterior segment exam were clear OU. MR was diagnosed with OMA. Upon examination of other family members, it was found that OMA is present in three consecutive generations. However, the presentation varied with all patients due to variable degrees of severity and expressivity. All family members positive for OMA also have BSNHL.

DISCUSSION: OMA is not thought to be genetic; however, this case study indicates it is likely autosomal dominant with variable expressivity. Regardless of the underlying cause of OMA, it is important for the practitioner to be aware of the various ocular presentations and patient adaptations of OMA because it is often overlooked as a diagnosis in the routine exam.

Degree Type
Thesis

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Ocular Motor Apraxia: A Familial Case Report

By

Tawna Roberts

A thesis submitted to the faculty of the College of Optometry
Pacific University
Forest Grove, Oregon
for the degree of
Doctor of Optometry
May 2003

Advisor

J.P. Lowery, MEd., O.D.
Ocular Motor Apraxia: A Familial Case Report

Author: Tawna L. Roberts

Advisor: J.P. Lowery
Author Biography

Tawna Roberts is originally from Southern Oregon. She received a Bachelor’s of Science degree in Biology from Pacific University in 1999. She will graduate with a Doctorate of Optometry in May of 2003. She plans to pursue a career as a pediatric optometrist.
Acknowledgments

I would like to take this opportunity to thank Dr. J.P. Lowery. Thank you for inviting me to get involved in this patient's care and taking me on as a student researcher for this thesis. Thank you for all the time you put in on reading, and proofing and re-reading and re-proofing my thesis.
BACKGROUND: Ocular Motor Apraxia (OMA) is a congenital ocular-motor anomaly involving the intermittent inability to initiate horizontal saccades. Smooth pursuit movements are usually normal in these patients. Signs of OMA are most apparent during infancy. Compensatory behaviors include head thrusting, blinking, and tilted head posture. Previous studies have shown that most affected children are slow in attaining early developmental milestones, and later tend to be clumsy. Difficulties in speech were very common and reading difficulties are also very well recognized in this condition. OMA is considered rare but there are no prevalence studies. The literature has previously suggested that OMA is not a genetic anomaly.

CASE REPORT: Eight month old, MR, was brought in for a vision exam because her mother was worried of possible visual dysfunction. Only positive health history is bilateral sensory-neural hearing loss (BSNHL). Visual acuity was 20/100 OU with Preferential Looking Cards. Retinoscopy revealed low hyperopia OU and visual fields full OU. Fixation was equal and central OD and OS. Vertical saccades and pursuits are normal. Horizontal pursuits were normal following abnormal head thrusting due to lack of horizontal saccades. Anterior segment and posterior segment exam were clear OU. MR was diagnosed with OMA. Upon examination of other family members, it was found that OMA is present in three consecutive generations. However, the presentation varied with all patients due to variable degrees of severity and expressivity. All family members positive for OMA also have BSNHL.
DISCUSSION: OMA is not thought to be genetic; however, this case study indicates it is likely autosomal dominant with variable expressivity. Regardless of the underlying cause of OMA, it is important for the practitioner to be aware of the various ocular presentations and patient adaptations of OMA because it is often overlooked as a diagnosis in the routine exam.
INTRODUCTION

Ocular Motor Apraxia (OMA) was first reported by Cogan in 1952.\textsuperscript{1} He used the term OMA to describe four children who experienced difficulty with horizontal saccades, although vertical saccades were normal. OMA is a congenital oculomotor anomaly involving the intermittent inability to initiate saccades and a failure of quick phases during optokinetic nystagmus (OKN) and vestibular nystagmus (VN). However, smooth pursuit movements are usually normal in these patients.

OMA is considered to be a rare anomaly but there are no prevalence studies. However, familial cases have been reported.\textsuperscript{5,6,7,11} Signs of OMA are most apparent during infancy. Compensatory behavior to shift gaze direction include characteristic headthrusting, blinking and tilted head posture, which enables the use of vertical eye movements that are usually unaffected.\textsuperscript{9}

During the characteristic head thrust, the eyes are driven to the extreme opposite side if the orbit due to the vestibuloocular reflexes (VOR). The head movement extends beyond the target, dragging the eyes with it until they are aligned with the target; the head then moves slowly back, the eyes remaining fixated on the target again by using the VOR.\textsuperscript{2} Although head thrusting is considered to be the most common characteristic, a study done by Shawkat, et al have shown that as little as 57\% of patients had head thrusting.\textsuperscript{9} Young infants or children with developmental delay will not use headthrusting as a compensatory behavior. Results of that study show that reliance of head thrusting as a sign of OMA will lead to underdetection. However, the practitioner must be aware that headthrusting is not seen exclusively in OMA patients. Practitioners must also rule out gaze palsies, slow saccades, visual field defects and poor eccentric
Another compensatory behavior to achieve change in direction of fixation is synkinetic blinking, but, this is usually seen in the older child or when the head is restrained.

Children with OMA have a failure of the quick phase during nystagmus. This can easily be examined in office by manually spinning the child or infant. While spinning, the slow phase drives the eyes to the extreme gaze. Without the quick phase, the eyes remain fixed, or ‘locked-up’. When children are young, looking for lock-up will aide in the diagnosis of OMA. However, lock-up is not reliable in infants prior to one month of age. Also, it is shown that there is a significant decrease in LU with age which is a normal maturation process with age even in patients with normal saccadic function.

It is important for OMA to be detected early by practitioners. A study done by Harris et al showed that most affected children are slow in attaining early developmental milestones, and later they tend to have poor motor skills. Difficulties in speech were very common (87%), and reading difficulties are also very well recognized in this condition. These delays occur regardless of the underlying diagnosis, occurring in the idiopathic children as well. Early detection will allow the child treatment in eye movement therapy to lessen the degree of delay they may experience. However, long term, the possibility of mild/moderate educational difficulties should be recognized. From the practitioner’s standpoint; diagnosis becomes more difficult because the signs and symptoms decrease with age.
MR, a seven-month-old female, was brought into the Oregon School for the Blind on 5/29/01 because her mother and early educational specialists were concerned about her visual function. Her mother noticed MR turned her head to look at faces and other objects. MR’s ocular history was only positive for a tendency to turn or tilt her head. Medical history revealed the pregnancy for MR lasted for 40.6 weeks and her mother was diagnosed with gestational diabetes. An intrathecal anesthesia was administered during the birth and the delivery was unremarkable. Birth weight was seven pounds and fifteen ounces and was noted to be doing well immediately after birth. In MR’s first year of life she had been prescribed amoxicillin and gentamycin eye drops. MR has been diagnosed with Bilateral Sensory Neural Hearing Loss (BSNHL) which is transmitted via an autosomal dominant pattern in her family.

MR’s distance visual acuity was 20/100 with the Preferential Looking Cards at 40cm. Her refractive status was measured to be low hyperopia OU. It was noted that she possessed good peripheral responses for gross visual fields. Bruckner and Hirshberg tests revealed equal and central fixation OU. It was also noted that she would use head thrusting to initiate horizontal eye movements ninety percent of the time. Her vertical eye movements were normal for both saccades and pursuits.

MR had little to no response to OKN testing. The Denver Developmental Profile revealed MR to be delayed in language, as expected, and possibly have a mild delay in gross motor. Her pupils were normal with a negative afferent papillary defect. Cycloplegic refraction revealed +2.50 diopters sphere OU. Anterior and posterior segment health was unremarkable.
MR was diagnosed with Hereditary Oculomotor Apraxia. Ocular and Denver Developmental Profile testing showed no evidence of neuro-pathology except for the expected delays in language due to the BSNHL.

During the initial history, MR’s father was observed using abnormal head movements while reading. This observation led to the initial diagnosis of OMA and further investigation into a possible genetic component of OMA. Family history indicated that other family members had possible eye movement dysfunctions with a possible genetic linkage between OMA and BSNHL. It was found upon examination that MR’s father, aunt, and her paternal grandmother demonstrate oculomotor patterns suggestive of the adult form of OMA. Also tested were three of MR’s cousins and her sister. It was found that one cousin definitely had OMA and OMA is probable for another cousin. All of the associated family members also have BSNHL.

MR’s father’s extra-ocular movements are full OU. He has a right hyperphoria on adduction and vertical diplopia in left gaze likely due to a right superior oblique underaction. Her father also showed vertical saccades with a horizontal correction. MR’s paternal grandmother is the first family member with a diagnosis of BSNHL. She had full movement in extra-ocular muscle testing. Cover test revealed alternating small angle esotropia (ten prism diopters). It did not appear as though she had OMA but it cannot be ruled out due to the variable degree and expressivity of the anomaly. All of MR’s family members who do not have the BSNHL also did not have any signs of OMA. However, all of the associated family members who had probable signs of OMA also have BSNHL (Figure 1). Two cousins who were positive for BSNHL appeared upon
examination to be OMA negative. However, due to the varying degrees of OMA and the presentation of OMA as children begin adolescents, it can not be completely ruled out.

DISCUSSION

OMA is not thought to be a genetic disorder, yet there have been documented cases. However, the particular family examined in this case has OMA phenotypically expressed in two and possibly three consecutive generations. Therefore, OMA is likely to be AD with variable expression in this family. All of the family members in this particular family that express OMA also have BSNHL. However, there were two individuals with BSNHL who do not show clinical signs of OMA. Therefore, linkage of OMA and BSNHL is unlikely.

As patients get older, they learn to adapt to this disorder by numerous behaviors; head tilt, blinking, and/or abnormal head movements. All of these compensatory head mechanisms are used by one or more of the family members examined in this study.

Early detection is key for children with OMA. Not only due to potential delay in reaching developmental milestones, but also most patients experience learning difficulty as they reach school age. Parents should encourage their children to make horizontal eye movements without undergoing head thrusting. This can be done by gently holding the child’s head to inhibit the head thrusts or by putting a beanbag on the older child’s head. These children also need to be seen frequently by an optometrist to monitor their progress with their eye movements.
Figure 1.

Black: (+) Sensory Neural Hearing Loss
Red: (+) Ocular Motor Apraxia, (+) Sensory Neural Hearing Loss
Green: Probable (-) Ocular Motor Hearing Loss, (+) Sensory Neural Hearing Loss
WORKS CITED


