Case report

Horizontal gaze palsy with progressive scoliosis in a Moroccan family

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A B S T R A C T

Horizontal gaze palsy with progressive scoliosis (HGPPS) is a rare clinical condition characterized by a combination of horizontal gaze palsy, pendular nystagmus and scoliosis. Only a few cases have been previously described in the literature. Our observations serve to document the first cases in Morocco.

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1. Introduction

Horizontal gaze palsy with progressive scoliosis (HGPPS) is a rare clinical condition [1] with autosomal recessive inheritance [2,3]. It is characterized by the combination of scoliosis, horizontal gaze palsy and pendular nystagmus [1,4,5]. There are few very published case reports. Here we report on the first Moroccan cases from a family affected by the condition and then review the literature on this rare clinical entity.

2. Patients and methods

An 11-month-old female infant from a non-blood related marriage, being cared for in orthopaedics for scoliosis, was referred by her family doctor to ophthalmology for treatment of nystagmus. Clinical examination found a low-amplitude, horizontal pendular nystagmus associated with horizontal gaze palsy, but with convergence and vertical eye movements intact.

Upon discussion with the parents, we discovered that three other siblings also had scoliosis. Examination of two brothers and one sister found the same ophthalmologic signs described above (Fig. 1) associated with head tremor during gaze fixation attempts. The infant is now 8 years old, wears a corset and continues to receive orthopaedic care for her scoliosis (Fig. 2). Two brothers (currently 16 and 22 years old) refused any treatment for their scoliosis (Fig. 3). The sister (currently 25 years old) was operated at age 15 for scoliosis.

3. Results

Review of literature on combined horizontal gaze palsy and scoliosis led to the diagnosis of HGPPS, which in turn led us to perform a neuroradiology evaluation in our patients.

CT scan of the brain and cervical, thoracic and lumbar spine found kyphoscoliosis and revealed that the floor of the fourth ventricle was tent-shaped in all patients and the bulbomedullary junction in two patients had a butterfly appearance. MRI was only performed on one patient, because of high cost. This provided more information on the previously described malformations (Figs. 4, 5).

In terms of ophthalmology, the brothers and oldest sister had bilateral amblyopia with corrected visual acuity of 4/10 in both eyes, while the younger sister had visual acuity of 8/10 in both eyes.

4. Discussion

HGPPS is an autosomal recessive inherited condition [2,3] secondary to mutation of the ROBO3 gene on chromosome 11 [3,6]. This gene encodes a protein participating in the decussation process of the motor and sensory pathways during the development of the central nervous system [6]. This mutation impedes the decussation process [3,7].

This paper reports on the first Moroccans affected by this condition. Our cases had the same abnormal findings as those described in the published literature, namely scoliosis with horizontal gaze palsy, with convergence and vertical eye movement intact but with pendular nystagmus [1,4,5]. Head tremor during gaze fixation attempts is not a consistent finding [5].
Neuroradiology evaluations based on CT scan and preferably MRI will help to reveal the malformations typical in patients with this condition. Some of the abnormal findings associated with this syndrome are the tent shape of the floor of the fourth ventricle, butterfly appearance of the bulbomedullary junction, reduced anteroposterior diameter of the pons and spinal cord [5,8] and lack of facial colliculus [8]. The lack of facial colliculus suggests selective agenesis of the nucleus of the sixth pair of cranial nerves, which explains the horizontal gaze palsy [8]. The lesions found in the cerebral trunk may explain the scoliosis, as demonstrated in studies performed on rats [7,9]. Functional MRI, diffusion tensor imaging and fibre tractography along with neurophysiology investigations help to confirm the lack of decussation of motor and sensory pathways [10].

CT scans performed on our patients provided evidence of the tent shape of the floor of the fourth ventricle in all patients and butterfly appearance of the bulbomedullary junction in two patients. The additional MRI examination in one of the patients provided further information.

HGPPS is a rare clinical condition that is not well known by either ophthalmologists or orthopaedic surgeons. Since scoliosis is the most striking outward sign, the ophthalmologic symptoms are often relegated to a position of secondary importance. This leads to delayed care for the nystagmus and any potential ametropia. The delay in ophthalmologic treatment in three of our patients resulted in bilateral amblyopia, which reduced the corrected visual acuity to
4/10 in both eyes. Because the young female patient treated by the paediatric surgeon wore appropriate optical correction and rehabilitation for amblyopia was implemented early on, her corrected visual acuity was 8/10. The goal of ophthalmologic care in patients presenting with HGPPS is to restrain the amblyopia to achieve better visual acuity despite the known oculomotor disorders. Based on this fact, we emphasize the need for early diagnosis of this condition and for collaboration between paediatric ophthalmologists and orthopaedic surgeons to ensure adequate care of patient with HGPPS syndrome.

5. Conclusion

The HGPPS cases described here are the first ones reported in Morocco and provide an opportunity for genetic research on this syndrome in the Moroccan population.

Disclosure of interest

The authors declare that they have no conflicts of interest concerning this article.

References