MR imaging of familial superior oblique hypoplasia

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ABSTRACT
Background Congenital superior oblique palsy is usually associated with a structural abnormality of the superior oblique tendon. There have been many reports of familial congenital superior oblique palsy. However, there has been no MRI documentation of familial superior oblique hypoplasia.
Methods Ophthalmological examination and orbital MRI were performed in three patients in a pedigree with familial superior oblique palsy. They showed typical signs of superior oblique palsy, including superior oblique underaction and overelevation in adduction on the affected side, torticollis in the early part of life, and positive head tilt testing.
Results Moderate to severe superior oblique hypoplasia was identified in all three affected family members.
Conclusion Superior oblique hypoplasia confirmed with MRI was useful for clarifying the aetiology of familial superior oblique palsy.

Congenital superior oblique palsy is usually associated with a structural abnormality of the superior oblique tendon.¹⁻⁴ There have been many reports of familial congenital superior oblique palsy.⁵⁻⁷ However, there has been no MRI documentation of familial superior oblique hypoplasia. We discovered a pedigree with three affected family members, who were 11 months, 7 years, and 27 years of age, respectively. The purpose of this study was to document the familial occurrence of superior oblique hypoplasia for the first time.

PATIENTS AND METHODS
Ophthalmological examination and orbital MRI were performed in three patients (11 months, 7 years and 27 years of age, respectively) with familial superior oblique palsy in a pedigree (figure 1). All of them showed typical signs of superior oblique palsy, including superior oblique underaction and inferior oblique overaction on the affected side, torticollis in the early part of life, positive head tilt test. Ophthalmological examinations were performed by JMH, and the evaluation of MRI by JHK.

MRI was performed on a 3 T system (Intera Achieva; Philips Healthcare, Best, The Netherlands) to evaluate the extraocular muscles in the orbits. Thin-section orbital T2-weighted imaging was performed in the orthogonal coronal plane with a turbo spin-echo technique to evaluate the extraocular muscles, using the following parameters: repetition time/echo time 3657/120 ms, field of view 150×150 mm, matrix 256×256 and section thickness 2 mm. Additionally, axial T2-weighted imaging was performed with 2 mm section thickness to cover the orbit in one patient, and 4 or 5 mm section thickness to cover the entire brain and orbit in the other two patients.

To determine hypoplasia of the muscles, the right and left extraocular muscles were compared based on a side-by-side visual evaluation for their size and shape along the whole length on the coronal T2-weighted images.

RESULTS
Moderate to severe hypoplasia of the superior oblique at the whole length including the tendon and belly was identified in all three affected family members.

Case 1
An 11-month-old boy presented with a head tilt to the right, which had been noticed at 2–3 months of age. Past medical history was not significant. The patient fixed and followed well with both eyes. He had a small left hyperphoria in the primary position, which increased with head tilt to the left. He also showed overelevation in adduction (figure 2A) and left superior oblique underaction. Coronal and axial T2-weighted images of the orbit showed hypoplasia of the left superior oblique at the whole length including the tendon and belly (figure 2B–E). All other extraocular muscles were symmetrical in size and shape.

Figure 1 Family pedigree. Affection status is as follows: darkened circles or squares denote an affected individual; clear denotes unaffected. Squares indicate male; circles indicate female; diagonal line indicates deceased.
His mother, a 27-year-old woman, showed orthotropia at distance and near in every gaze and with the head tilt to either side. Her ductions and versions were normal.

**Case 2**
A 7-year-old boy, a maternal cousin of Case 1, presented with head tilt to the left, first noticed around 1 year of age. Past medical history was not significant. He had right hypertropia of 12 prism dioptres (PD) at distance and right exotropia of 10 PD and hypertropia of 18 PD at near in the primary position without correction. His right hypertropia was 6 PD to the right gaze, and 14 PD to the left gaze, increased to 25 PD with head tilt to the right and decreased to 4 PD with head tilt to the left. The patient exhibited right overelevation in adduction and right superior oblique underaction (figure 3A). He also showed facial asymmetry with a fuller right side of face. Coronal and axial T2-weighted images of the orbit showed hypoplasia of the right superior oblique at the whole length including the tendon and belly (figure 3B–E). All other extraocular muscles were symmetrical in size and shape.

**Case 3**
A 27-year-old woman, the mother of Case 2, presented with reduced but persistent head tilt to the left from early childhood, even though she underwent a strabismus surgery at a local clinic 2 years prior. She had right exotropia of 14 PD and right hypertropia of 6 PD in the primary position, right exotropia of 14 PD and right hypertropia of 6 PD in the right gaze, and right hypertropia of 6 PD in the right gaze, and right hypertropia of 6 PD in the right gaze.

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**Figure 2** Superior oblique palsy in Case 1. (A) Ocular versions demonstrating inferior oblique overaction in the left eye. (B–E) Two-millimetre-section coronal T2-weighted images showing hypoplasia of the left superior oblique at the tendon (B) and belly (C, D) (arrows). On the 4-mm-section axial T2-weighted image (E), the right superior oblique (arrows) is well visualised, but the left superior oblique is not.
exotropia of 14 PD and right hypertropia of 8 PD in the left gaze. With the head tilt to the right, she showed right exotropia of 16 PD and right hypertropia of 6 PD, and with head tilt to the left, right exotropia of 16 PD and no hypertropia. She showed right superior oblique underaction (figure 4A) and facial asymmetry with a fuller right side of the face. Coronal and axial T2-weighted images of the orbit showed severe hypoplasia of the right superior oblique (more severe at the belly portion) (figure 4B–E). All other extraocular muscles were symmetrical in size and shape.

Figure 3  Superior oblique palsy in Case 2. (A) Ocular versions demonstrating inferior oblique overaction and superior oblique underaction in the right eye. (B–E) Two-millimetre-section coronal T2-weighted images showing hypoplasia of the right superior oblique at the tendon (B) and belly (C, D) (arrows). On the 5-mm-section axial T2-weighted image (E), the left superior oblique (arrows) is clearly visualised, but the right superior oblique is not.
DISCUSSION
MR imaging documentation of familial superior oblique hypoplasia has not been previously described. In this study, moderate to severe superior oblique hypoplasia was identified in all three affected family members. They showed typical signs of superior oblique palsy, including superior oblique underaction and inferior oblique overaction on the affected side, torticollis in the early part of life, and positive head tilt testing. Even though this study included only three patients, superior oblique hypoplasia might at least partly explain the aetiology of previously reported cases.

Figure 4  Superior oblique palsy in Case 3. (A) Ocular versions demonstrating superior oblique underaction in the right eye. (B–E) Two-millimetre-section coronal T2-weighted images showing severe hypoplasia of the right superior oblique at the tendon (B) (arrows). The belly portion of the right superior oblique is not found (C, D), suggesting more severe atrophy. On the 2-mm-section axial T2-weighted image (E), the left superior oblique (arrows) is clearly visualised, but the right superior oblique is not.
Oblique muscle palsy and were suggested as one of the risk factors. Morphisms were found in the patients with congenital superior fibrosis of the extraocular muscles. Even though our patients did not show the typical recessive pattern, a further study to investigate the presence of ARIX gene polymorphisms might be helpful to understand the pathophysiology of familial congenital superior oblique palsy. In conclusion, MR imaging proved useful for clarifying the aetiology of familial superior oblique palsy.

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**Competing interests** None.

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