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CASE REPORT

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Unexpected extraocular muscle hypoplasia during strabismus surgery: case series

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Abstract

Background Reports of congenital isolated medial rectus muscle abnormalities are relatively uncommon and are seldom seen. According to our clinical experience, some rare cases of abnormalities could not be detected by clinical examination and imaging before surgical treatment, which brought difficulties to diagnosis and surgery.

Case presentation In order to provide clinical guidance, we summarized 4 cases with congenital hypoplasia of the medial rectus muscle in our hospital recently. All the patients exhibited exotropia in the primary position. Only one patient (25.0%) exhibited clinically significant limitations of ocular movements. All the patients were identified with congenital hypoplasia of the medial rectus muscle during strabismus surgery; one patient also had hypoplasia of the lateral rectus muscle. However, abnormalities of the rectus muscles were not identified by MRI in three patients (75.0%). In terms of treatment, we enhanced the surgery amount in three patients. Good correction of exotropia was achieved in all patients.

Conclusions Congenital hypoplasia of the medial rectus muscle is extremely rare and some cases are difficult to be detected by clinical examination or imaging. Surgeons should be aware of this condition and should actively but cautiously adjust the surgical parameters based on the patients' intraoperative status.

Keywords Extraocular muscle, Congenital abnormality, Strabismus surgery

Background

Congenital hypoplasia of the extraocular muscles (EOMs) is rare. Although there have been multiple reports of the absence of inferior rectus (IR), superior rectus (SR), or superior oblique muscles [1–3], there have been few reports of congenital abnormalities of the medial rectus (MR) or lateral rectus (LR) muscles. Previous reports have indicated that the common features of such abnormalities include large-angle strabismus and substantially limited ocular movement. Kiarudi et al [2] described one patient who lacked a right MR muscle and exhibited no adduction in the right eye. Murthy et al [4] reported that a patient who lacked MR muscles in both eyes showed large-angle exotropia and the absence of adduction beyond the midline in both eyes.

Nevertheless, based on our clinical experience, irregularities cannot be identified through clinical examination

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and imaging in some rare cases, making it difficult to diagnose before surgical treatment. To our knowledge, such rare cases have not been reported previously. Therefore, we conducted this study to describe four patients with congenital hypoplasia of the MR muscle; one patient also had hypoplasia of the LR muscle. Our findings may serve as clinical guidance for surgeons to pay closer attention to this muscle hypoplasia condition and make active yet cautious adjustments to the surgical plan during the surgery.

Case presentation

Methods

The medical records of four patients with congenital hypoplasia of the EOMs subjected to surgical correction at Department of Ophthalmology and Visual Science, Eye and ENT Hospital of Shanghai Medical College, Fudan University, Shanghai, China, between June 2020 and June 2022 were retrospectively reviewed. The clinical features including age, gender, ocular alignment, ocular motility, intraoperative findings, modified surgical plans, postoperative imaging, muscle biopsy and final outcomes were analyzed. Patients with dystrophia or absence of the EOMs were included in our study. All patients were examined and treated by one doctor (CZ). Age, gender, family history, cycloplegic refraction, ocular motility, ocular alignment, intraoperative findings, preoperative and modified surgical plans, postoperative orbital imaging, final outcomes and follow-up were obtained from the patients' charts.

Good correction was defined by a horizontal deviation of 10 prism diopters (PD) or less and a vertical deviation of 5 PD or less in the primary gaze as assessed during the final follow-up visit. The last follow-up time ranged from 1 month to 2 years.

The research was conducted ethically in accordance with the World Medical Association Declaration of Helsinki. Ethics approval for this study was obtained from the Institutional Review Board of Eye and ENT Hospital, Fudan University. Written informed consent was obtained from the parents of all patients described in this report for both study participation and publication of identifying information/images in an online open-access publication.

Results

There were three males (75.0%) and one female (25.0%) included in our research. The mean age at surgery was 2.5 ± 0.5 years. Three patients (75.0%) had hyperopia, while one (25.0%) had myopia. All the patients presented with horizontal exotropia; the median degree was 55 (range = 40 to > 90) PD. Only one patient (25.0%) exhibited clinically significant limitations (−3 limitation on a scale of 0 to −4) [5] of ocular movements. Abnormal head position was observed in one patient (25.0%). Congenital hypoplasia of the MR muscles was revealed in all the patients during strabismus surgery, while one patient was also with hypoplasia of the LR muscle. We enhanced the surgery amount in three patients, and remained the surgery plan in one patient. Good correction of exotropia was achieved in all patients. Hypoplasia of the rectus muscles was not identified by postoperative MRI in three patients (75.0%). Main clinical data of the 4 cases were summarized in Table 1. All the cases are presented as follows.

Table 1 Main clinical data of 4 cases with congenital hypoplasia

Case No./ Age (y)/ sex	PACT (PD)	Horizonal motility limitation	Diagnosis	Previous surgery plan	Intraoperative findings	MRI findings	Modified surgery plan
No.1/2/F	60	−1	CXT, left IOOA	LMR + 5, LLR-8, RMR-8, LIO anterior transposition	Absence of RMR and RLR, LMR degeneration	All EOMs thin but exist	LMR + 8, LLR-9, LIO anterior transposition
No.2/3/M	50	−1	IXT, bilateral IOOA	LMR + 5, LLR-8, BIO tenotomy	LMR degeneration, LMR adheres to Tenon's capsule	No obvious abnormalities	LMR + 5, LLR-8, BIO tenotomy
No.3/3/M	> 90	−3	CXT, left IOOA	BMR + 7, BLR-9, LIO anterior transposition	BMR degeneration	No obvious abnormalities	BMR + 9, BLR-9, LIO anterior transposition
No.4/2/M	40	0	CXT, right SOOA, right Brown Syndrome	RMR + 5, RLR-6.5, RSO nasal tenotomy	Absence of RMR, thinness of other EOMs	Absence of RMR	LLR-7, RLR-6.5, RSO tenotomy

Y year, *F* Female, *M* Male, *PD* Prism diopter, *CXT* Constant exotropia, *IOOA* Inferior oblique overaction, *IXT* Intermittent exotropia, *SOOA* Superior oblique overaction, *L* Left, *R* Right, *B* Bilateral, + Resection, — Recession, *MR* Medial rectus, *LR* Lateral rectus, *IO* Inferior oblique, *SO* Superior oblique, *EOM* Extraocular muscle

Case 1

Medical history and ocular findings

A 2-year-old girl presented with left exotropia that had begun at the age of 6 months. There was no family history of strabismus. The patient was healthy with no history of systemic disease. Cycloplegic refraction revealed a value of + 1.75 DS in the right eye and + 2.25 DS in the left eye. She had 60 PD of left exotropia, as determined by the prism alternating cover test (PACT) in the primary position (Fig. 1A). Ocular movement examination indicated a mild adduction limitation (−1 limitation on a scale of 0 to −4) in the right eye, along with overaction of the left inferior oblique (Fig. 1A). Fundus and ultrasound examinations showed no abnormalities. The patient was diagnosed with constant exotropia with left inferior oblique overaction (IOOA); she underwent corrective surgery in both eyes.

Preoperative surgical plan

Forced duction test (FDT), left MR resection 5 mm, left LR recession 8 mm, right LR recession 8 mm, and left inferior oblique (LIO) anterior transposition.

Surgical procedures

FDT revealed mild restrictions of adduction and abduction in the right eye, and posterior adhesions were suspected. Intraoperative exploration revealed that the MR and LR muscles in the right eye could not be reached with strabismus forceps (Fig. 1B). Dystrophy of the MR muscle was observed in the left eye. In order to correct the horizontal strabismus, we increased the surgical quantity of the horizontal muscles in the left eye. The surgical plan was modified as follows: left MR muscle resection 8 mm, left LR recession 9 mm, and LIO anterior transposition.

Outcome and follow-up

Postoperative MRI scans showed bilateral orbital asymmetry and slight thinning of all EOMs. However, all EOMs were present, including the right MR and LR muscles (Fig. 1C). Biopsy of the left MR muscle revealed collagen and vascular tissue with scattered inflammation (Fig. 1D), indicating degeneration of the left MR muscle. The postoperative diagnosis was modified to non-concomitant exotropia, left IOOA, left MR muscle degeneration, and right EOM displacement.

Postoperative follow-up

At 1 month after surgery, the patient was orthophoric in primary gaze and with right IO overaction. (Fig. 1E).

Case 2

Medical history and ocular findings

A 3-year-old boy presented with right exotropia that had been present since birth. There was a history of forceps use at birth. Cycloplegic refraction revealed a value of + 1.75 DS in the right eye and + 1.50 DS in the left eye.

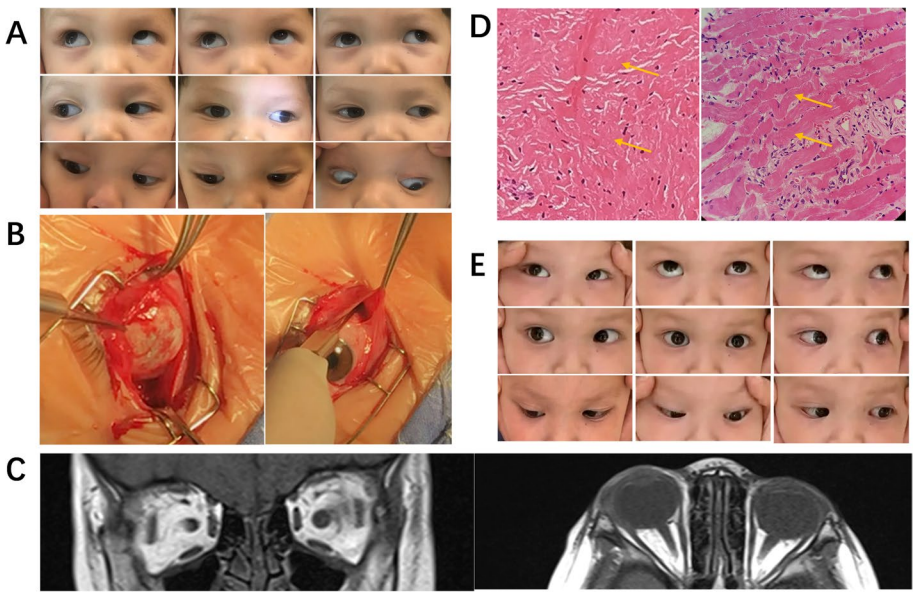

Fig. 1 Case 1: A 2-year-old girl with hypoplasia of the right MR and IR muscles. **A** Preoperative picture showing 60 PD of left exotropia in the primary position. **B** Surgeon's view showing absence of the LR (left) and MR (right) muscles in the right eye. **C** Postoperative MRI scans showed bilateral orbital asymmetry and slight thinning of all EOMs. **D** Biopsy of the left MR muscle (left) revealed collagen and vascular tissue with scattered inflammation (left arrows) compared with normal muscle (right arrows). **E** At 1 month after surgery, the patient showed little horizontal deviation (< 5 PD) in the primary position but with right IO overaction

He had 50 PD right exotropia, as determined by PACT in the primary position (Fig. 2A). Ocular movement examination indicated a mild adduction limitation (−1 limitation on a scale of 0 to −4) in both eyes, along with overaction of the bilateral inferior obliques (Fig. 2A). Fundus and ultrasound examinations showed no abnormalities. The patient was diagnosed with intermittent exotropia (IXT) with bilateral IOOA; he underwent corrective surgery in both eyes.

Preoperative surgical plan

FDT, left MR resection 5 mm, left LR recession 8 mm, and bilateral inferior oblique tenotomy.

Surgical procedures

Intraoperative exploration revealed dystrophy of the MR muscle, which resembled connective tissue, in the left eye (Fig. 2B). Furthermore, the left MR muscle was closely adhered to the Tenon's capsule. Although dystrophy of the MR muscle might reduce its force generation, its adhesion to surrounding tissues could potentially enhance the force exerted. Therefore, ultimately, we did not alter the surgical plan. The surgical plan remained as planned: left MR resection 5 mm, left LR recession 8 mm, and bilateral inferior oblique tenotomy.

Outcome and follow-up

Postoperative MRI scans did not show any obvious abnormalities (Fig. 2C). However, biopsy of the left MR muscle revealed collagen and vascular tissue with scattered inflammation, indicating degeneration of the left MR muscle.

Postoperative follow-up

At 1 month after surgery, the patient showed no heterotropia in the primary position, and his ocular movement was normal. During 1 year of follow-up, the patient showed 5 PD right exotropia in the primary position. His ocular alignment remained stable (Fig. 2D).

Case 3

Medical history and ocular findings

A 3-year-old boy presented with left exotropia, which had been present since the age of 3 months. There was no family history of strabismus. Cycloplegic refraction revealed a value of +0.00 DS in the right eye and −0.25 DS in the left eye. He had a large-angle exotropia (> 90 PD), as determined by PACT in the primary position (Fig. 3A). In both eyes, ocular movement examination revealed deficits of adduction beyond the midline (−3 limitation on a scale of 0 to −4) during elevation. Fundus photography revealed large cups in both eyes (Fig. 3B), while intraocular pressure monitoring and glaucoma screening did not reveal any abnormalities. Ultrasound examinations showed no abnormalities. The patient was diagnosed with constant exotropia, left IOOA and a physiological large cup-to-disc ratio; he underwent corrective surgery in both eyes.

Preoperative surgical plan

FDT, bilateral MR resection 7 mm, bilateral LR recession 9 mm, LIO anterior transposition.

Surgical procedures

During surgery, abnormal development of the MR muscles was observed in both eyes. The bilateral MR muscles resembled connective tissue and the muscle fibers were thin (Fig. 3C). Notably, normal muscle fibers were

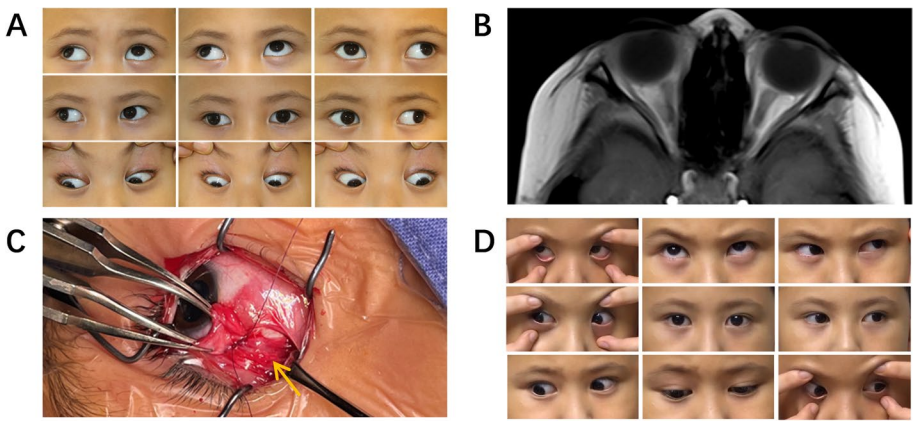

Fig. 2 Case 2: A 3-year-old boy with hypoplasia of the left MR muscle. **A** Preoperative picture showing 50 PD of right exotropia in the primary position. **B** Surgeon's view showing dystrophy of the MR muscle (arrow), which resembled connective tissue, in the left eye. **C** Postoperative MRI scans did not show any obvious abnormalities. **D** During 1 year of follow-up, the patient showed 5 PD right exotropia in the primary position

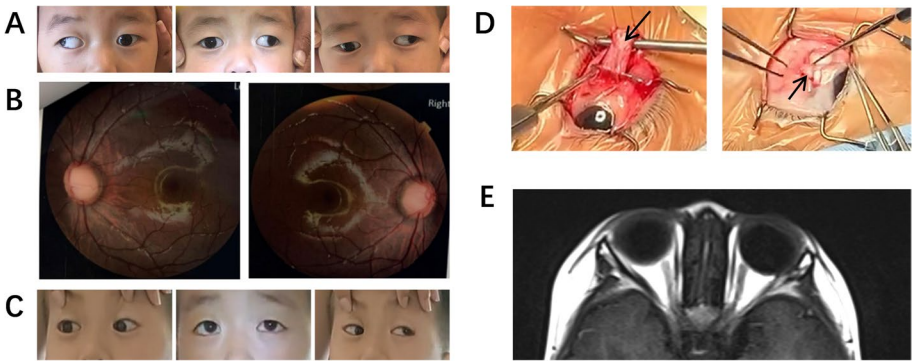

Fig. 3 Case 3: A 3-year-old boy with hypoplasia of MR muscles in both eyes. **A** Preoperative picture showing a large-angle (> 90 PD) of left exotropia in the primary position. Three gaze pictures revealed deficits of adduction beyond the midline (−3 limitation on a scale of 0 to −4) during elevation in both eyes. **B** Fundus photography revealed large cups in both eyes. **C** Muscle fibers of the left MR muscle (left arrow) were thin. Surgeon's view showing degeneration of the MR muscle (right arrow), which resembled connective tissue, in the right eye. **D** Postoperative MRI scans did not show any obvious abnormalities of the orbits or EOMs. **E** Good correction of exotropia was confirmed at the 2-year follow-up

present 1 cm posterior to the bilateral MR muscles. Considering the partial dystrophy of the bilateral MR muscles, which might lead to a decrease in muscle elasticity, the surgical quantity was increased. The surgical plan was modified as follows: bilateral MR resection 9 mm, bilateral LR recession 9 mm, LIO anterior transposition.

Outcome and follow-up

Postoperative MRI scans did not show any obvious abnormalities of the orbits or EOMs (Fig. 3D). Biopsy of the bilateral MR muscles revealed collagen fibers and vascular adipose tissue with scattered inflammation, indicating degeneration of the bilateral MR muscles.

Postoperative follow-up

At 1 month after surgery, the patient showed no horizontal deviation in the primary position and no underaction in any direction of gaze. Good correction of exotropia was confirmed at the 2-year follow-up (Fig. 3E).

Case 4

Medical history and ocular findings

A 2-year-old boy presented with right exotropia (Fig. 4A) and a clinically significant left face turn (Fig. 4B). There was no family history of strabismus. The patient was healthy with no history of systemic disease. Cycloplegic refraction revealed a value of + 1.25 DS in the right eye and + 2.25 DS in the left eye. He had 40 PD right exotropia and 5 PD right hypertropia as determined by PACT in the primary position. Fundus and ultrasound examinations showed no abnormalities. Ocular movement examination indicated that the right eye did not elevate in adduction, and overaction of the right superior oblique muscle was observed. The patient was diagnosed with constant exotropia, along with superior oblique overaction (SOOA) and Brown syndrome in the right eye; he underwent corrective surgery in both eyes.

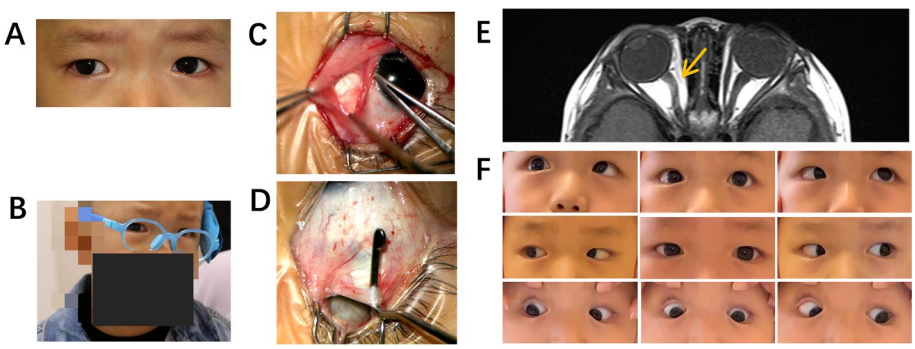

Fig. 4 Case 4: A 2-year-old boy with hypoplasia of the right MR muscle. **A** Preoperative picture showing 40 PD of right exotropia and 5 PD right hypertropia in the primary position. **B** The patient presented with a clinically significant left face turn. **C**, **D** Surgeon's view showing absence of MR muscle (**C**), while other EOMs, such as the SO muscle (**D**), were thin in the right eye. **E** Postoperative MRI scans revealed the absence of the right MR muscle(arrow). **F** At 8 months after surgery, the patient demonstrated orthophoria in the primary position, along with vertical alignment abnormalities in upgaze

Preoperative surgical plan

FDT, right MR resection 5 mm, right LR recession 6.5 mm, and right superior oblique tenotomy on the nasal side.

Surgical procedures

FDT revealed a mild restriction of gaze toward the superior-medial position in the right eye. Intraoperative exploration revealed that the MR muscle in the right eye could not be reached with surgical forceps (Fig. 4C), while other EOMs were thin (Fig. 4D). Because of these intraoperative findings, the surgical plan was modified as follows: left LR recession 7 mm, right LR recession 6.5 mm, and right superior oblique tenotomy.

Outcome and follow-up

Postoperative MRI scans revealed the absence of the right MR muscle (Fig. 4E).

Postoperative follow-up

Eight months post-surgery, the patient exhibited orthophoria in the primary position, along with vertical alignment irregularities during upward gaze. (Fig. 4F).

Discussion and conclusions

Congenital abnormalities of EOMs have previously been reported in the literature [1–4, 6, 7]. The most frequent abnormalities involve the superior oblique muscle [7], followed by the IR muscle [8]. Congenital absence or dysplasia of the SR, MR, and LR muscles are extremely rare [4, 9, 10]. In this paper, we described four patients with congenital hypoplasia of the MR muscle, with or without the LR muscle; these abnormalities were not detected by clinical examinations or postoperative imaging. An important aspect of our report is that we observed various degrees of limitation of ocular movement, in contrast to the findings in previous reports. In addition, we identified discrepancies between imaging results and intraoperative findings. Both of these aspects led to difficulties of making an accurate diagnosis before surgery.

The pathogenesis of EOM abnormalities remains unknown. EOMs and their tendons appear to be derived from a superior mesenchymal complex and an inferior mesenchymal complex. The MR and LR muscles develop from both complexes during embryological development [11]. Damage during this stage of development may cause congenital abnormalities of the EOMs.

Previous reports regarding congenital abnormalities of the horizontal rectus muscles generally mention large-angle strabismus and limited ocular movement. Kiarudi et al [2] described a 37-year-old man with absence of the MR muscle in the right eye; his clinical features included > 90 PD exotropia and the absence of adduction in the right eye. Murthy et al [4] described a 19-year-old man who lacked MR muscles in both eyes; he exhibited large-angle exotropia and the absence of adduction beyond the midline in both eyes. Although all of our patients exhibited exotropia in the primary position, they had various degrees of limitation of their ocular movements. The patient in case 3 exhibited substantial limitation of ocular movement, similar to the findings in previous reports. In contrast, the patient in case 1 had hypoplasia of the MR and IR muscles in the right eye, but exhibited only mild limitation of ocular movement. The patient in case 2 had degeneration of the left MR muscle, although normal adduction was observed in both eyes. One possible explanation for this finding is that backward EOM displacement afforded some movement compensation, while the SR muscles maintained some inward and outward rotation functionality.

Imaging techniques are considered essential for the objective evaluation of EOM anomalies [3, 4, 12]. Zhu et al [13] reported that routine imaging was effective for identifying any probable structural changes in EOMs. Therefore, information provided by EOM imaging can support definitive decisions concerning surgical management [14]. However, postoperative MRI scans of our patients did not reveal any obvious EOM abnormalities, except in the patient in case 4. This discrepancy between imaging results and intraoperative findings has also been reported in the literature [2, 10]. Sharma et al [10] explained this discrepancy in terms of the embryological development of EOMs. EOMs consist of two types of muscle fibers (global and orbital), which develop separately during embryological development. The global fiber layer extends over the entire length of the muscle, while the orbital fiber layer terminates at the equator of the globe. Therefore, when only the orbital fiber layer is present, muscles can be detected by imaging posterior to the equator, while they cannot be detected anterior to the equator during surgery. This explains why normal muscle fibers were observed 1 cm posterior to the bilateral MR muscles in the patient in case 3. This may also explain why ocular movement was not substantially limited in the patients in cases 1 and 2—only the global fiber layer was missing, while the orbital fiber layer was present.

Regardless of the reason, in our patients, congenital hypoplasia of the MR muscle could not be diagnosed before surgery, which hindered proper surgical planning. MR muscle abnormalities were identified by intraoperative direct observation, and biopsies of the muscles

provided further diagnostic guidance. FDTs were performed to determine the amount of restriction [15]; these tests may also provide etiological insights regarding congenital abnormalities of the MR muscles.

Due to the inconformity of muscle morphology inconformity between preoperative MRI findings and intraoperative observation, the surgical plans for strabismus management were modified. Considering the elasticity of dystrophic muscles is different from that of normal muscles, often a larger surgical quantity is required to achieve the desired effect. Therefore, we chose to enhance the surgery amount in three of the patients (case 1, 3, 4). Fu et al [6] also recommended initial overcorrection surgery for strabismus caused by congenital dysplasia of EOMs; this approach resulted in a low rate of postoperative recurrence and led to satisfactory long-term surgical outcomes. However, in case 2, after considering the quantitative relationship between muscle degeneration and the amount of muscle adhesion, we did not modify the surgical plan. Therefore, we suggest that when encountering such rare events, comprehensive consideration is necessary, and the method of modifying the surgical plan should be carefully considered. Good correction of exotropia was achieved in all patients.

There are several limitations in this case report. Since all the patients were from one hospital and diagnosed by one doctor, the results cannot be generalized. Besides, since the congenital abnormalities of EOMs in our patients were not observed until surgery, we did not prepare a CCD microscope camera. Instead, photos during the operation were taken by mobile phones, which may have resulted in the resolution may be low.

In conclusion, we have described four patients with congenital hypoplasia of the MR muscle, with or without the LR muscle; these abnormalities were diagnosed during strabismus surgery. All the patients exhibited exotropia in the primary position, while only one patient exhibited clinically significant limitations of ocular movements. Hypoplasia of the rectus muscles was not identified by MRI in three of the four patients. The surgical plans were re-discussed during surgery. We enhanced the surgery amount in three patients, and all patients achieved good correction of exotropia. Our findings provide insights into rare abnormalities and may help surgeons reconsider surgical approaches based on the patients' intraoperative status.

Abbreviations

EOM	Extraocular muscle
FDT	Forced duction test
IOOA	Left inferior oblique overaction
IR	Inferior rectus
LIO	Left inferior oblique
LR	Lateral rectus
MR	Medial rectus
MRI	Magnetic resonance imaging
PACT	Prism alternating cover test
PD	Prism diopters
SR	Superior rectus

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Authors' contributions
YQY was the primary contributor in writing the manuscript. LL and WW collected the patient data and were major contributors in writing the manuscript. XYW, XBY and CJ enrolled patients into the study, contributed to the analysis and interpretation of the patient data. CZ and WW revised it critically for important intellectual content. All authors read and approved the final manuscript.

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Data availability
No datasets were generated or analysed during the current study.

Declarations

Ethics approval and consent to participate
The research was conducted ethically in accordance with the World Medical Association Declaration of Helsinki. Ethics approval for this study was obtained from the Institutional Review Board of Eye and ENT Hospital, Fudan University. Written informed consent was obtained from the parents of all patients described in this report for the publication of their clinical findings.

Consent for publication
Written informed consent was obtained from the parents of all patients described in this report for both study participation and publication of identifying information/images in an online open-access publication.

Competing interests
The authors declare no competing interests.

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