

Congenital dysplasia involving both medial and inferior recti: clinical features and surgical outcomes

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Abstract

• **AIM:** To investigate the clinical features and surgical outcomes of congenital dysplasia involving both inferior recti (IR) and medial recti (MR) muscles.

• **METHODS:** A retrospective review was conducted including cases of simultaneous congenital dysplasia of IR and MR that were diagnosed and surgically treated at the Zhongshan Ophthalmic Center, Sun Yat-sen University, China, from July 2009 to November 2019. Ocular motility, ocular alignment at distance (6 m) and near (33 cm) by prism alternating cover test and stereoacuity were assessed in all patients before and after surgery.

• **RESULTS:** A total of five patients (four males and one female; three with right eye and two with left eye congenital dysplasia) were included in this review. The patients ranged in age from 10 to 42y (21±13.4y). The main clinical findings were hypertropia and exotropia of the affected eye, along with motility limitations in adduction and depression. Lateral rectus (LR) recession/transposition combined with IR resection was performed in one case. Two scheduled surgeries were performed in four cases, with one involving superior rectus recession and IR resection and the others LR recession and MR resection. Mean±SD pre-surgical exotropia of 51.0±31.11 prism diopter (PD) and hypertropia of 29.20±7.12 PD in the primary position were decreased to 3.6±12.90 and 3.2±10.09 PD, respectively, at two years after surgery, with a success rate of 60% and an under-correction rate of 40%.

• **CONCLUSION:** The main clinical features associated with simultaneous MR/IR congenital dysplasia are hypertropia and exotropia of the affected eye along with motility limitations in adduction and depression. Scheduled two-stage surgeries achieved a success rate of 60%.

• **KEYWORDS:** medial rectus; inferior rectus; paralytic strabismus; strabismus surgery; dysplasia of extraocular muscle

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INTRODUCTION

Congenital dysplasia of extraocular muscles, as can result from embryonic mesodermal lesions of unknown origins, represents a rare congenital anomaly. Although congenital dysplasias or isolated absences of inferior recti (IR) and medial recti (MR) muscles have been reported in the literature^[1-6], to our knowledge, no paper on congenital dysplasia simultaneously involving MR and IR muscles currently exists. Thacker *et al*^[7] reviewed four cases with simultaneous MR and IR paresis; however, all of these cases were complicated due to endoscopic sinus surgery. Jiao *et al*^[8] analyzed 13 cases of oculomotor nerve palsy, three of which presented with simultaneous MR and IR paresis and showed satisfactory outcomes after surgical treatment. However, again, these cases did not involve a simultaneous congenital dysplasia of MR and IR muscles. Therefore, given the complete absence of information on this condition, it is imperative to relate the clinical characteristics and surgical outcomes associated with this rare disease. In this report, we review five cases of simultaneous congenital dysplasia involving both MR and IR muscles which were confirmed by preoperative orbital magnetic resonance imaging (MRI) or computed tomography (CT) examinations and postoperative histopathological evaluation. The clinical features and surgical management of these cases are discussed.

SUBJECTS AND METHODS

Ethical Approval The study was performed according to the tenets of the Declaration of Helsinki and was approved by the Research Ethics Board of the Zhongshan Ophthalmic Center (No.2019KYPJ103) of Sun Yat-sen University, China. Written informed consent to participate in this study was obtained from all patients or from the parents of those younger than 18y.

A retrospective analysis of patients who had a simultaneous congenital dysplasia of MR and IR muscles and who were seen and treated at the Zhongshan Ophthalmic Center of Sun Yat-sen University, China, between July 2009 and November 2019 was conducted. All patients were surgically treated by the corresponding author (Yan JH).

The diagnostic criteria for inclusion in the study included: 1) a positive non-progressive exotropia and hypertropia of the affected eye in the primary position since birth; 2) significant motility limitations in adduction and depression of the affected eye; 3) negative forced duction test results as performed under general anesthesia; 4) a diagnosis that had been confirmed by preoperative orbital CT or MRI and postoperative histopathological examination. The exclusion criteria were not having received strabismus surgery and not followed up within the previous six months or more.

The following data were recorded: age, sex, affected eye, family history, age at onset, best corrected visual acuity, cycloplegic refraction, anterior segment fundus photograph, ocular motility, ocular alignment at distance (6 m) and near (33 cm) by prism alternating cover tests in nine diagnostic positions, and stereoacuity at distance and near. Cyclotropia was measured from fundus photographs (obtained by recording the fovea position as related to the optic disc) and/or with use of double Maddox rod testing. All patients underwent orbital MRI and/or CT examinations. Limitations in eye movement in all diagnostic gazes were measured using a 5-point scale (0 to -4) as described previously^[9].

All surgeries were performed through fornix conjunctival incisions while patients were under general anesthesia. The forced duction test was routinely performed during surgery. In these five patients, a total of nine surgeries of various types were performed in all affected eyes. Lateral rectus (LR) recession/transposition combined with IR resection was performed in one case while planned two-stage surgeries were performed in the remaining four cases. The two-stage surgeries consisted of superior rectus (SR) recession (7-8 mm) and IR resection (8-10 mm) performed to correct vertical strabismus, followed six months later with LR recession (7-10 mm) and MR resection (7-10 mm) to correct horizontal strabismus. Patients were followed up at one day, one week, two months, six months, one year, and two years after surgery. A complete cure was defined as a vertical deviation of ≤ 5 PD and a horizontal deviation of ≤ 10 PD in the primary position as determined on their final visit. Failures were defined as residual deviations of >6 PD in vertical deviation and >11 PD in horizontal deviation^[10-12]. Statistical analysis was performed using the SPSS version 23.0 program (SPSS Inc, Chicago, IL, USA). All values are presented as mean \pm standard deviations.

RESULTS

A total of five patients (four males and one female) were included in this review, with their age being 21 ± 13.4 (10-42)y. The right eye was affected in three patients (60%) and the left in two (40%). The age at onset was 1.8 ± 0.3 y, with a long-preoperative time of 19.8 ± 13 (8-40)y. Family history in all cases was not remarkable. Prior to surgery, none of the five cases experienced diplopia, nor did any of these cases show stereopsis or abnormal head position. One case showed optic nerve atrophy along with a trauma-induced medial orbital wall fracture; four cases had anisometropic refractive error; three cases had mixed strabismic and anisometropic amblyopia; two cases had congenital nystagmus; and one case presented with lagophthalmus.

Images from orbital CT/MRI scans were available for all five cases with CT scans alone in three cases, MRI alone in one case, and both MRI and CT examinations in one case. Orbital MRI/CT scans showed extreme thinning or atrophy of both MR and IR muscle bellies in all five patients (Figure 1C). In one patient with a history of trauma, MRI revealed a medial orbital wall fracture that did not affect ocular alignment and ocular motility, as no changes in his ocular motility and strabismus were present after injury.

Prior to surgery, the overall exotropia in primary position at distance was 51 ± 31.11 PD, with measurements in four patients ranging from 30 to 45 PD and in one patient 105 PD. Hypertropia in primary position at distance was 29.20 ± 7.12 PD. Limitations of duction in MR and IR gazes were -3.2 ± 0.45 and -3.6 ± 0.55 , respectively. The final postoperative exotropia in primary position at distance was 3.6 ± 12.90 PD; Hypertropia in primary position at distance was 3.2 ± 10.09 ; and limitations of duction in MR and IR gazes were -1.2 ± 0.45 and -1.4 ± 0.55 , respectively (Table 1).

Nine types of surgery based on the angle of deviation and residual function of affected muscles were performed in these five cases. LR recession and MR resection were performed to correct horizontal strabismus in one case (case 1), while SR recession and IR resection were performed to correct vertical strabismus in four cases (cases 2, 3, 4, and 5). LR recession and 1/2 tendon width downward transposition combined with IR resection was performed in one case (case 1). At the time of surgery, the affected MR and IR muscles were found to be very thin and relaxed. There were no abnormal fibrous bands within or around the other operated extraocular muscles. Patients were followed up for 8 ± 8.4 (6 to 34)mo. Four cases showed a postoperative exotropia of ≤ 10 PD, and one case had a residual exotropia of 25 PD in the primary position as determined on their final visit. Three cases had postoperative hypertropia of ≤ 5 PD, and two cases had a residual hypertropia of >10 PD.

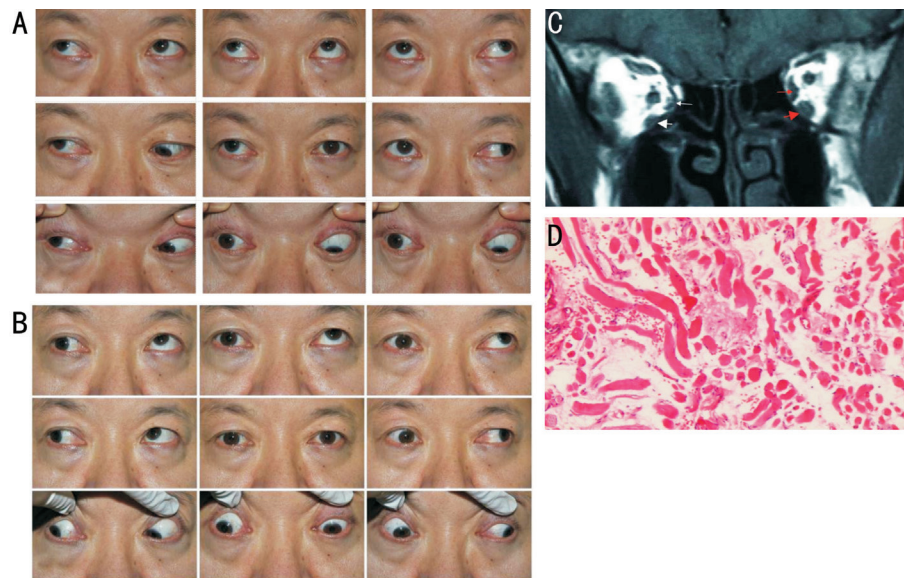


Figure 1 Male, aged 42 (case 2) with simultaneous congenital dysplasia involving both medial and inferior recti muscles (OD) A: Before surgery, this patient had 40 PD right hypertropia and 35 PD exotropia in the primary position and -4 underaction in both down and left gazes of the right eye. B: Six months after the second stage of this two-staged strabismus surgery, he showed 4 PD hypotropia of the right eye in the primary position and -1 and -2 underaction in down and left gazes, respectively. The first surgery consisted of a 7 mm superior rectus recession and 10 mm inferior rectus resection of the right eye, while the second was 7 mm lateral rectus recession and 7 mm medial rectus resection of the right eye. C: Orbital MRI coronal scan revealed that the size and cross-sectional area of the right inferior rectus (thick white arrow) and medial rectus (thin white arrow) were smaller and thinner than those of the left inferior rectus (thick red arrow) and medial rectus (thin red arrow). D: Pathological examination of the resected inferior rectus revealed a substantial amount of derangement and atrophy of striated muscles and a marked increase in the amount of connective fibrous tissue (HE×40).

Table 1 Pre- and post-operative deviations and motility in simultaneous congenital dysplasia involving both medial and inferior recti muscles

Pat./sex/ age (y)/eye	Pre-deviation (PD) (horizontal, vertical)	Motility limitation		Surgical procedures	Post-deviation (PD) (horizontal, vertical)	Motility limitation	
		MR	IR			MR	IR
1.F/15/R	RXT45, RHT25	-3	-4	RLR Rec10 mm/transp (1/2)/RIR Res 12 mm	RET8, RHT5	-1	-1
2.M/42/R	RXT35, RHT40	-4	-4	RSR Rec 7 mm/RIR Res10 mm; RLR Rec 7 mm/RMR Res7 mm	RXT4, 0	-2	-1
3.M/27/L	LXT105, LHT30	-3	-3	LLR Rec10 mm/LMR Res10 mm/LSO tenotomy; LSR Rec 8 mm/LIR Res 8 mm	LXT5, RHT10	-1	-2
4.M/12/R	RXT30, RHT30	-3	-3	RSR Rec 8 mm/RMR Res 8 mm; RSR Rec 3 mm/RIR Res 8 mm	RXT25, RHT18	-1	-2
5.M/13/L	LXT45, LHT21	-3	-4	LSR Rec7 mm/LIR Res 8 mm; LMR Res 9 mm/LLR Rec 10 mm	0, LHT3	-1	-1

Pat: Patient; F: Female; M: Male; R: Right; L: Left; PD: Prism diopter; HT: Hypertropia; RET: Right esotropia; LET: Left esotropia; RXT: Right exotropia; LXT: Left exotropia; RHT: Right hypertropia; LHT: Left hypertropia; SO: Superior oblique; SR: Superior rectus; IR: Inferior rectus; MR: Medial rectus; LR: Lateral rectus; Rec: Recession; Res: Resection; LSO: Left superior oblique; Transp: Transposition.

Fusion function, anisometropia, and the best corrected visual acuity significantly improved in two cases, and stereopsis function recovered in one patient at one year after strabismus surgery. Overall, outcomes were successful in three patients (cases 1, 2, and 5) when assessed at their final follow-up (Figure 1A and 1B), while the remaining two patients were under-corrected, but showed a substantial improvement in their appearance. No other complications such as anterior segment ischemia syndrome or any new in- or ex-cyclotropia were present in these patients.

Pathological examinations of resected IR and MR muscles revealed a substantial amount of derangement in striated

muscles and a small amount of connective fibrous tissue. These findings contributed to the validation of extraocular muscle dysplasia diagnosis (Figure 1D).

DISCUSSION

Strabismus related to congenital abnormalities of extraocular muscles have been reported in previous literatures^[6,13-14]. Simultaneous congenital dysplasia involving both MR and IR muscles is extremely rare. Its diagnosis is based on findings of hypertropia and exotropia of the affected eye with motility limitations in adduction and depression and negative forced duction test results. In addition, in the present study, orbital CT/MRI scans revealed that both MR and IR showed extreme

thinning and atrophy. Pathologically, the resected IR and MR muscles demonstrated a substantial amount of derangement of striated muscles and a small amount of connective fibrous tissue, which further confirms the presence of muscle dysplasia. The pathogenesis of this maldevelopment in MR and IR muscles remains unclear. It is possible that traumatic, inflammatory, metabolic, toxic, and/or hypoxic events exerted at the site of medial and inferior rectus nuclei/fascicles or terminal branches of the oculomotor nerve serving these two recti occurring at birth or at perinatal stages may lead to such a congenital dysplasia. However, none of our patients reported any history of these events. In all five cases, the palsy was monocular and sporadic, which differs from the few cases with dysplasia or absence of isolated extraocular muscles that are binocular and familial^[15-17]. Due to the early onset of strabismus and amblyopia, none of our cases showed abnormal head positions, nor did they experience diplopia.

While this condition could be confused with a partial cranial nerve III palsy and muscle atrophy^[18], patients with the latter condition often show other clinical features, such as ptosis, dilated pupil, and lack of accommodation^[8]. Another potential entity in the differential diagnosis is either iatrogenic or traumatic damage/rupture of both MR and IR muscles. However, traumatic damage to MR and IR muscles typically involves a history of local injury, often accompanied by sudden diplopia, periorbital ecchymosis, proptosis, retrobulbar hemorrhage, and orbital blow-out fracture^[7]. Based on our findings and experience, we emphasize the diagnostic value of orbital CT or MRI examinations in cases of traumatic orbital fracture and undefined damage of extraocular muscles, including both congenital and acquired conditions. Finally, the observation of other acquired paralytic strabismus effects upon both MR and IR muscles, such as myasthenia, chronic progressive external ophthalmoplegia, and idiopathic orbital myositis, can be easily differentiated by their own clinical presentations.

All our cases underwent strabismus surgery. Clearly, neither SR/IR horizontal transposition nor MR/LR vertical transposition may be used for strabismus correction when both MR and IR muscles are paralyzed^[7,19]. Therefore, LR recession and MR resection were performed to correct horizontal strabismus in one case, while SR recession and IR resection were performed to correct vertical strabismus by two planned surgeries in the remaining four cases. In staging surgeries, we recommend that the vertical strabismus should be corrected first, followed by horizontal strabismus correction in the second surgery 6-12mo later, to minimize risk associated with surgery on multiple muscles on the same occasion. However, to avoid a second surgery in patients with limited or small deviations, a rectus transposition was performed in

one of our patients. In this case, a LR recession and downward transposition were combined with IR resection with the result that a satisfactory alignment was achieved within a single surgery. This surgical technique was similar to that reported by Jiao *et al*^[8] involving a MR resection combined with one-half to two-thirds of a tendon width of superior transposition being performed as a means to simultaneously correct the exotropia and hypotropia in cases with oculomotor nerve palsy.

As the MR and IR muscles were paretic in our patients, enhanced surgeries involving high degrees of both rectus recession and resection were required in order to balance the extreme disparities of forces exerted upon the palsied eye. In this study, the amounts of recession or resection of rectus muscle was 8-12 mm, and the dose-effect relationship was slightly larger than previously reported^[8,12,20-22]. The presence of both MR and IR palsy, along with the contracture of LR and SR muscles in the affected eye, makes a postoperative recurrence almost certain. Therefore, early surgical overcorrection is necessary. We recommend a mild overcorrection in the initial postoperative period, which will result in a low rate of postoperative recurrence and enable a long-term satisfactory surgical outcome. Jiao *et al*^[8] suggested an overcorrection of 5°-10° esotropia during surgery for partial oculomotor nerve palsy and 10°-15° esotropia for total oculomotor nerve palsy. Gesite-de Leon *et al*^[20] suggest that an initial overcorrection of 15 PD in visually mature cases should be considered, as the findings of their study indicated an average late exoshift of 17 PD after MR advancement, while Donaldson *et al*^[21] found that a small-angle overcorrection of 5 to 10 PD for exotropia enabled a suitable ocular alignment immediately after surgery. Our experience suggests that an initial overcorrection has better outcomes for both vertical and horizontal strabismus. For cases with preoperative vertical deviations of >20 PD, a suitable overcorrection was 10 to 15 PD, and for vertical deviations from 10 to 20 PD, a suitable overcorrected was 5 to 10 PD. For horizontal deviations of 20 to 40 PD, an appropriate overcorrection was 10 to 15 PD, and for horizontal deviations of >45 PD, it was 15 to 20 PD. Overcorrections remained at one day, one week and one month after surgery for both vertical and horizontal strabismus, while orthophoria or heterophoria was found at one to two years after surgery, in accordance with previous reports^[12,21]

The limitations associated with this study include the retrospective nature of the case review and limited case sample size. As patients not receiving strabismus surgery were excluded, this may introduce sampling bias. In addition, detailed cerebral imaging was not performed in our series as no associated brain problems were suspected in these patients. Finally, longer follow-up times along with larger sample sizes will be needed in future studies.

In conclusion, simultaneous congenital dysplasia involving both IR and MR muscles is extremely rare. The main clinical features include hypertropia and exotropia of the affected eye, motility limitations in adduction and depression, and negative forced duction test results. Surgical procedures include LR recession/transposition and IR resection for mild cases, while in severe cases, SR recession and IR resection in the first stage, followed by LR recession, and MR resection in the second-stage surgery are recommended. Suitable overcorrection in the initial postoperative period results in a satisfactory outcome with a success rate of 60%. Further investigations into the etiology of this condition, as can be achieved with detailed neuroimaging and genetic analyses, is required.

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