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Self-Harm Traumatic Cataract and Vitreous Haemorrhage in a Girl with Marden-Walker Syndrome – A Case Report

Traumatska katarakta i krvarenje u staklovinu nastalo samoozljeđivanjem kod djevojke s Marden-Walkerovim sindromom – prikaz slučaja

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Abstract. ***Aim:*** The aim of this case report is to highlight the clinical presentation, diagnosis, and treatment of a young Marden-Walker syndrome (MWS) patient who developed a traumatic cataract and vitreous haemorrhage. It underscores the importance of multidisciplinary awareness in managing MWS, advocating for timely ophthalmologic assessments, especially after ocular trauma in patients with complex genetic syndromes. ***Case report*****:** A 20-year-old female with MWS and intellectual disability was referred to the Department of Ophthalmology of University Hospital Centre Zagreb due to a white opacity in her right pupil from repetitive self-harm. Her medical history included craniofacial abnormalities, hypotonia, joint contractures, and self-injurious behaviour, particularly frequent hitting and rubbing the eye. A dense, mature traumatic cataract in her right eye was identified. An ultrasound of the eye was performed preoperatively, and vitreous haemorrhage was established. Due to the lack of cooperation, visual tests were limited, and surgery was performed under general anaesthesia, involving lens aspiration, anterior vitrectomy and intraocular lens implantation. The patient was discharged the next day with the prescription of neomycin, polymyxin B, and dexamethasone combination for local use (eye drops 5x/day and ointment 2x/day). Besides that, isotonic eyewash solution containing potassium iodide was also prescribed for local use (eye drops 4x/day). At the follow-up, her vision had improved, with no complications. Multidisciplinary care helped reduce her self-harm, and no further ophthalmological interventions were needed. ***Conclusions*****:** This case report highlights the importance of early diagnosis and surgical treatment of traumatic cataracts in patients with MWS, as well as the need for psychological and ophthalmologic interventions to prevent complications. A multidisciplinary approach, involving genetics, psychiatry, and ophthalmology, is crucial for providing comprehensive care to this vulnerable group of patients.

Keywords: Cataract; Traumatic; Genetic Diseases; Inborn; Marden-Walker Syndrome; Patient Care Team; Self-Injurious Behaviour; Vitreous Haemorrhage

Sažetak. ***Cilj*****:** Cilj ovog prikaza slučaja jest naglasiti kliničku sliku, dijagnozu i liječenje mlade pacijentice s Marden-Walkerovim sindromom (MWS) koja je razvila traumatsku kataraktu i krvarenje u staklastom tijelu. Naglašava važnost multidisciplinarne svijesti u upravljanju MWS-om zagovarajući pravovremene oftalmološke procjene, osobito nakon traume oka kod pacijenata sa složenim genetskim sindromima. ***Prikaz slučaja*****:** 20-godišnja djevojka s MWS-om i intelektualnim teškoćama upućena je na Kliniku za oftalmologiju Kliničkog bolničkog centra Zagreb zbog bijelog zamućenja u desnoj zjenici, nastalog uslijed ponavljanog samoozljeđivanja. Njezina povijest bolesti uključivala je kraniofacijalne abnormalnosti, hipotoniju, kontrakture zglobova i samoozljeđujuće ponašanje, osobito često udaranje i trljanje očiju. Na pregledu se ustanovi gusta, zrela traumatska katarakta na desnom oku, zbog čega se učini i ultrazvuk oka te se ustanovi krvarenje u staklovinu. Zbog nedostatka suradnje, sam pregled bio je ograničen, a operacija učinjena u općoj anesteziji. Prilikom operativnog

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zahvata učinjena je aspiracija leće, prednja vitrektomija i implantacija intraokularne leće. Bolesnica je otpuštena idućeg dana uz propisanu kombinaciju lijekova neomicina, polimiksina B i deksametazona za lokalnu primjenu (kapi za oči pet puta dnevno i mast dva puta dnevno). Osim toga, propisana je i izotonična otopina za ispiranje očiju s kalijevim jodidom za lokalnu primjenu (kapi za oči četiri puta dnevno). Poslijeoperacijski tijek protekao je bez komplikacija, uz značajno poboljšanje vida prema anamnezi. Multidisciplinarna skrb pomogla je smanjiti njezino samoozljeđivanje i nisu bile potrebne daljnje oftalmološke intervencije. ***Zaključci*:** Ovaj prikaz slučaja naglašava važnost rane dijagnoze i kirurškog liječenja traumatske katarakte i krvarenja u staklastom tijelu u bolesnika s MWS-om, kao i potrebu za psihološkim i oftalmološkim intervencijama kako bi se spriječile komplikacije. Multidisciplinarni pristup, koji uključuje genetičare, psihijatre i oftalmologe, ključan je za pružanje sveobuhvatne skrbi ovoj ranjivoj skupini pacijenata.

Ključne riječi: autodestruktivno ponašanje; prirođene genetske bolesti; traumatska katarakta; krvarenje u staklovinu; Marden-Walkerov sindrom; multidisciplinarni tim za skrb o bolesniku

Marden-Walker syndrome (MWS) is an extremely rare congenital disorder. It is characterized by craniofacial abnormalities, arthrogryposis (multiple joint contractures), and severe developmental delays. The genetic cause of MWS remains unclear, though some patients have shown pathological variants in PIEZO2. The syndrome is likely inherited in an autosomal recessive manner.

INTRODUCTION

Marden-Walker syndrome (MWS) is an exceedingly rare congenital disorder first identified by Marden and Walker in 1966[1]. Syndrome is characterized by a distinctive triad of craniofacial dysmorphism, arthrogryposis (multiple joint contractures), and profound developmental delays[2–4]. The genetic aetiology of MWS has not yet been clearly established (pathological variants in PIEZO2 (18p11.22-p11.21) have been identified in some patients)[5–7]. MWS is likely inherited in an autosomal recessive pattern[8].

The incidence of MWS is currently unknown due to its rarity, and it is often underdiagnosed or misdiagnosed due to phenotypic overlap with other syndromes. To date, around 50 cases have been described in the world literature[3].

Clinically, individuals with MWS present with a spectrum of manifestations, including micrognathia, high-arched palate, blepharophimosis, a characteristic mask-like facies, and ocular abnormalities such as ptosis and strabismus. The musculoskeletal system is often markedly affected, with generalized hypotonia, joint contractures, and occasionally, pterygium formation. Neurological involvement includes varying degrees of intellectual disability and speech delays. Despite these established features, the full phenotypic spectrum of MWS remains incompletely characterized, particularly concerning ophthalmologic manifestations[4].

Ocular involvement in MWS primarily includes structural abnormalities like ptosis and strabismus. However, the development of cataracts, particularly secondary to trauma, and vitreous haemorrhage, are notably rare and underreported. Traumatic cataracts are typically associated with direct or indirect ocular injuries, resulting in lens opacity. In patients with MWS, the structural integrity of ocular tissues may be compromised due to underlying genetic abnormalities, potentially increasing susceptibility to trauma-induced cataracts. This is particularly concerning given that visual impairments can exacerbate developmental delays in this already vulnerable population[9].

The rarity of traumatic cataracts and vitreous haemorrhage in MWS poses significant challenges in clinical management and prognosis. Early diagnosis and intervention are crucial to prevent long-term visual impairment and associated developmental consequences. However, due to the rarity of MWS and the atypical presentation of ocular trauma, there is a paucity of literature guiding optimal management strategies.

This case report aims to fill this gap by detailing the clinical presentation, diagnostic workup, and treatment course of a young patient with MWS who developed a traumatic cataract and a vitreous haemorrhage. Consequently, it emphasizes the need for heightened awareness and multidisciplinarity among clinicians regarding the diverse manifestations of MWS to ensure timely and appropriate interventions, e.g. comprehensive ophthalmologic assessment in patients with complex genetic syndromes like MWS, particularly following any form of ocular trauma.

CASE REPORT

A 20-year-old female patient with a previously known diagnosis of Marden-Walker syndrome interconnected with intellectual disability was referred to the Ophthalmology Clinic of the University Hospital Centre Zagreb due to white opacity of her right pupil because of repetitive self-harm. The patient's medical history was significant for all the characteristic features of the syndrome, including distinctive craniofacial abnormalities, hypotonia, joint contractures, kyphoscoliosis, and developmental delay (Figure 1). Her caregivers noted a history of self-injurious behaviours, particularly frequent eye hitting and rubbing which had escalated over the past few months (Figure 2).

Bearing in mind the medical history and physical examination data, a dense, mature traumatic cataract of the right eye was identified at the tertiary level and the diagnosis of self-inflicted traumatic cataract was finally set (Figure 3). Because of the dense, mature traumatic cataract, preoperatively an ultrasound of the eye was performed and vitreous haemorrhage was found (Figure 4).

Due to the lack of cooperation, visual acuity tests and biomicroscopic evaluations could not be performed. Via basic direct ophthalmoscopy, a normal, so-called "red reflex" was noticed. The surgical procedure under general anaesthesia was suggested as the treatment standard[10]. The surgical procedure was performed and it consisted of lens aspiration, primary posterior capsulorhexis, anterior vitrectomy, and primary intraocular lens implantation, respectively. Since there were no early postoperative complications, the patient was discharged from the hospital the very next day with the prescription of neomycin, polymyxin B, and dexamethasone combination for local use (eye drops 5x/day and ointment 2x/day). Besides that, isotonic eyewash solution containing potassium iodide was also prescribed for local use (eye drops 4x/day), because of the vitreous haemorrhage. Postoperative care included visual rehabilitation and protective strategies to prevent further injury. At the follow-up visit three weeks post-surgery, the patient demonstrated a substantial improvement in visual acuity (as per

Figure 1. Clinical appearance and visible joint contractures

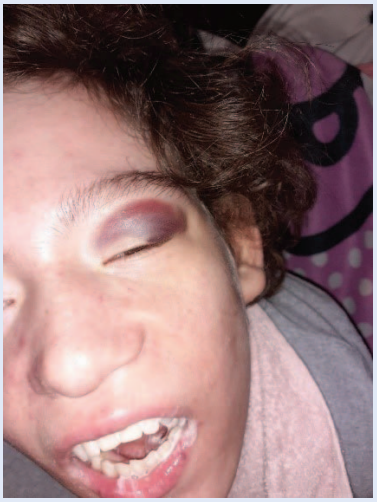

Figure 2. Self injury of the eye

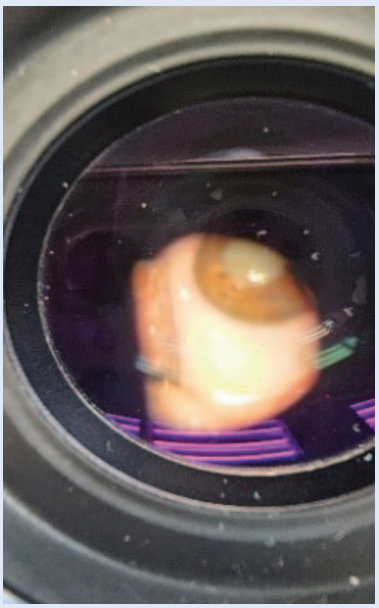

Figure 3. Traumatic mature cataract

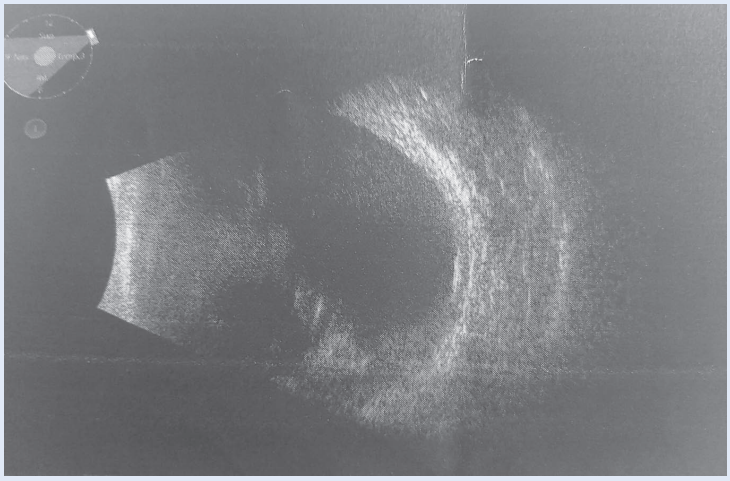

Figure 4. Vitreous hemorrhage on ultrasound

anamnesis) in the right eye, and no late postoperative complications were identified. Vitreous haemorrhage was also resorbed.

After ophthalmological cure, the patient was referred to the leading paediatric consultant (neuro-paediatric consultant and geneticist) as well as to psychiatry consultant and psychologist to work on/prevent behavioural tendencies that may lead to self-harm. To the best of our knowledge, following the targeted multidisciplinary approach, the pattern of self-harm was notably reduced and up to the present data, the patient did not need novel ophthalmological referrals nor procedures.

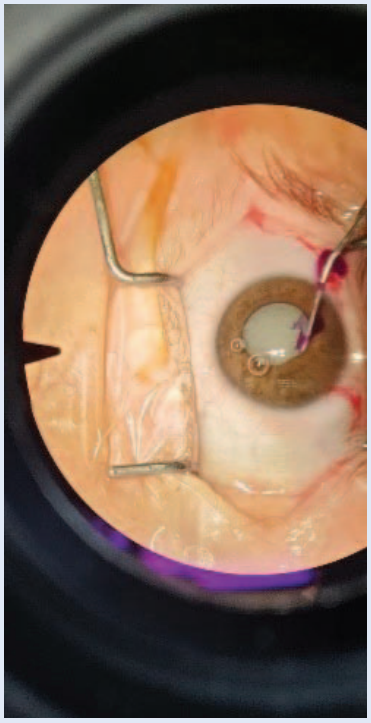

Figure 5. Traumatic mature cataract and gentiana violet for easier capsulorexis

DISCUSSION

MWS is an extremely rare genetic disorder, so any new knowledge about the manifestations of the disease is useful. In our case report, we have shown a rare complication of this syndrome, traumatic cataract and vitreous haemorrhage of the eye. According to our knowledge, by reviewing the literature, there is no such case described so far with an ophthalmological complication in a person with Marden Walker syndrome. Several authors also wrote about ophthalmological complications in patients with mental retardation who received chorioretinal ablation as a result of self-injury, and in one patient self-enucleation was even described[11, 12]. In addition to traumatic cataracts and vitreous haemorrhage, such repeated blows to the eye can, in patients with mental retardation, lead to a number of other eye diseases such as chorioretinal ablation, hyphema, hyposphagma, angle recession and consequent glaucoma, choroidal rupture, iridodialysis and uveitis[9, 13, 14].

Ocular trauma is one of the leading causes of unilateral blindness in the world, and traumatic cataract is a frequent consequence of eye injuries in adults and children[15]. It is important to emphasize that traumatic cataracts have more serious consequences and more complex treatment than senile cataracts. The injury is rarely limited to the lens. Damage can also be present on the zonules, posterior capsule and posterior segment. Therefore, the success of the operation is challenging[16, 17]. Due to the impossibility of cooperation during the actual examination and surgical procedure, the operation was performed under general anaesthesia, using the colour purple (gentiana violet) to perform the capsulorhexis (Figure 5).

Despite the cataract, it was not necessary to use phacoemulsification, but the lens masses were aspirated. Considering the patient's age, a posterior capsulorexy and an anterior vitrectomy were performed, after which an intraocular lens was implanted in the posterior chamber. Fortunately, the vitreous haemorrhage resorbed, so no fur-

ther surgery was required. The vision improved significantly postoperatively, as did the quality of life of our patient. In further follow-up, there is a risk of dislocation of the intraocular lens in the early postoperative period, and in the late period of fibrosis of the posterior capsule of the lens, despite posterior capsulorhexis and anterior vitrectomy. There is also a risk of repeated self-injury of the eyes.

Individuals with genetic disorders have multisystemic, complex and special health needs[18]. It is important to emphasize that timely recognition and treatment of the psychological aspects of MWS is crucial in preventing self-harm and its consequences. An interdisciplinary team that includes geneticists, orthopaedists, ophthalmologists, psychologists, and social workers can provide the comprehensive care necessary for these patients.

Family support plays a key role in the care of patients with MWS, especially when it comes to managing the psychological and physical challenges associated with the condition. Families are often the main providers of daily care and emotional support, which can significantly affect the outcome of treatment and the overall quality of life of patients. In the case of our patient, the continuous support of the family was crucial in recognizing the problem of self-harm and seeking appropriate medical help. Educating the family about the nature of MWS, its symptoms and possible complications enables them to better understand the condition and adequately react to changes in the patient's health. Also, families who are well informed about treatment options and available resources can effectively collaborate with medical teams in planning and implementing comprehensive care.

Psychosocial support within the family can significantly reduce stress and feelings of isolation in patients. Providing emotional support, encouraging positive behaviours and participating in therapeutic activities can help reduce the incidence of self-harm. In the case of our patient, family support was crucial in motivating her to participate in psychotherapy and other therapeutic procedures. In addition, family support may include organizing additional help and resources, such as joining support groups for families of patients with rare genetic disorders. These groups provide an opportunity to share experiences, advice and emotional support among families facing similar challenges. Environmental adaptation reduces the risk of self-harm by removing dangerous objects and providing a safe environment. Our case also points to the need for better strategies to manage self-harm in patients with rare genetic disorders. Psychotherapy, supported by pharmacological interventions, can play a key role in reducing the incidence of self-harm[18]. Regular ophthalmological examinations are also of crucial importance for the early detection and treatment of eye complications that may arise as a result of self-harm.

Individuals with genetic disorders have complex, multisystem health needs. Timely recognition and treatment of the psychological aspects of MWS are key to preventing self-harm. A multidisciplinary team, including geneticists, orthopaedists, ophthalmologists, psychologists, and social workers, is essential for providing comprehensive care.

CONCLUSIONS

With this case report, we wanted to show how repetitive eye trauma in a patient with Marden Walker syndrome can lead to traumatic cataracts, vitreous haemorrhage or any other ocular complication, and also to point out how the psychological changes due to this rare syndrome require close cooperation between different medical specialties to ensure optimal care and prevention for this vulnerable group of patients.

Conflicts of Interest: Authors declare no conflicts of interest.

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