CASE REPORT

# BILATERAL CONGENITAL MYDRIASIS WITH ACCOMPANYING MIXED DEVELOPMENTAL DELAY

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**ABSTRACT**

***Introduction:*** *Congenital bilateral mydriasis is a rare condition characterized by fixed dilated pupils unresponsive to light. It may occur associated with cardiovascular syndrome, gastrointestinal malformation, or developmental delay. Referred causes in comprehensive differential diagnosis for congenital mydriasis include alterations in ocular structures, ocular innervation alterations and others secondary to trauma, infection, or topical medication.*

***Case Report:*** *A 10 months-old boy presented to the hospital with large pupils in both eyes since birth. He has a persistent pupillary membrane with 9 mm diameter of pupil and unable to respond to light in both eyes. The pupillary response test using pilocarpine solution 0.1% showed no difference in diameter of pupils. Comprehensive pediatric examination revealed developmental delay. Computed tomography and laboratory result (laboratory blood count, toxoplasmosis, and cytomegalovirus) within normal limit. After eight months follow up, no significant change in pupil size or light responsiveness was found.*

***Discussion:*** *Comprehensive examination of anterior to posterior segments of the eyes is imperative on the lookout for underdiagnosed diseases. Clinical assessment remains vital for guiding investigations. Observation is required to detect the presence of certain systemic abnormalities that frequently do not manifest at birth.*

***Conclusion:*** *Congenital mydriasis can be diagnosed quickly in ordinary conditions, but it can be quite challenging when it presents as a coincidental finding of other spectrum diseases. It is important to perform a comprehensive general and eye examination to rule out any serious condition related to it.*

***Keywords:*** *Bilateral congenital mydriasis, dilated pupil, mixed developmental delay, pediatric ophthalmology*

## INTRODUCTION

Congenital mydriasis sometimes referred to as congenital fixed dilated pupil is a very rare eye condition. By definition, fixed and dilated pupils are present at birth and are found in otherwise normal eyes. The condition has been explained as a result of diminished muscle function, decreased cholinergic, sensitivity or production, and iris sphincter aplasia. This may be related to accommodation failure and associated with progressive neurological deterioration and smooth muscle cell (SMC) dysfunction. Neurological findings such as developmental delay should be investigated as there have been reports of this in previous cases congenital mydriasis. [1 2 3]

Delay in development is generally determined with a child whom does not attain developmental milestones as compared to peers of the same population.4 The etiology of the

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vast majority of developmental delay is idiopathic. When known, the cause may include genetic, environmental, and/or psychosocial factors. Systemic anomaly such as congenital heart defect should be investigated since there are reports about this in previous cases of congenital mydriasis pupils.5

This case report highlights the diagnostic complexities of bilateral congenital mydriasis accompanied by developmental delay, leading to notable physical signs and symptoms including changes of structure of the eyes by observation for 8 months.

## CASE ILUSTRATION

A 10-month-old boy was admitted to Hasanuddin University hospital with a chief complaint of large (dilated) pupils that are notable since the time of birth. From birth the mother noted dilated and unreactive pupils. The child was born at term, delivered through normal birth, weighed 3600 grams, born as the third child of the family. There are none precedents in family history nor any trauma in this case and was not related with any used medication.

Visual acuity was central, unsteady and unmaintained which means worse than 20/300. Both conjunctivas show a network of very fine blood vessels, with white underlying sclera and deep anterior chambers. There was persistent pupillary membrane in both eyes but irises remain within normal limits. Both pupils were round and regular but unresponsive to light. The size of both of the pupils were 9 mm in diameter. Intraocular pressure was within normal limits on each eye. In the presence of horizontal nystagmus, the cover test was challenging to evaluate, therefore Hirschberg test results were normal. Posterior segment examination revealed normal optic discs on both eyes, as well as general examination

In pupil examination by dripping pilocarpine 0.1% in both eyes, pupils were observed in two different rooms, each with different set of lightings (bright and dim), before and after instillation. The result showed that pupils did not respond to pilocarpine 0.1%, pupil diameter remained at 9mm in both eyes and was concluded to be unresponsive after 30 minutes of administration.

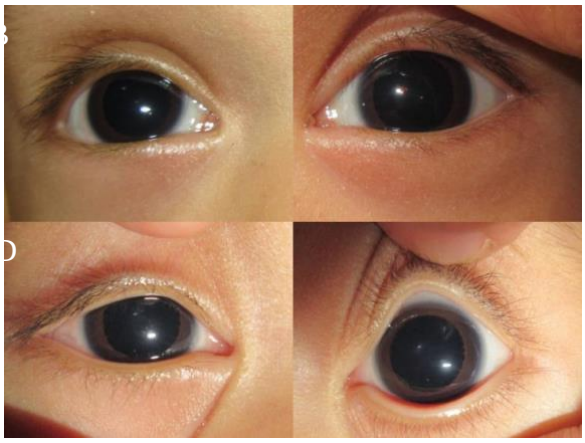

**Fig 1. Right and left pupil size before (A-B) and after pilocarpine 0,1% (C-D) in bright light room**

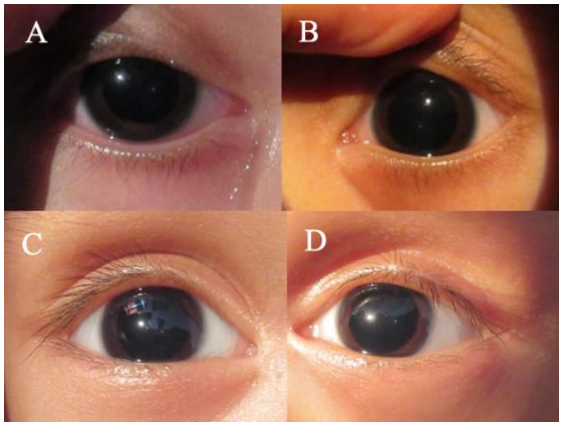


**Fig 2. Right and left pupil size 30 minutes before (A-B) and after pilocarpine 0,1%(C-D) in dimmed light room**

Additional examination was conducted found to looking for any neurological defects. Computed tomography with contrast of brain was shown within normal limit. Laboratory founding include torch and CMV are within normal limit. We referred to pediatric and cardiology unit. Pediatric examining suggested no other symptoms, defects, or any abnormalities has been found in this case. Furthermore, cardiology unit carried out echocardiography test and the result showed outcome within normal limit. This patient was diagnosed with bilateral mydriasis with a suspected of congenital anomaly with differential diagnosed to bilateral parese of ocular innervation.

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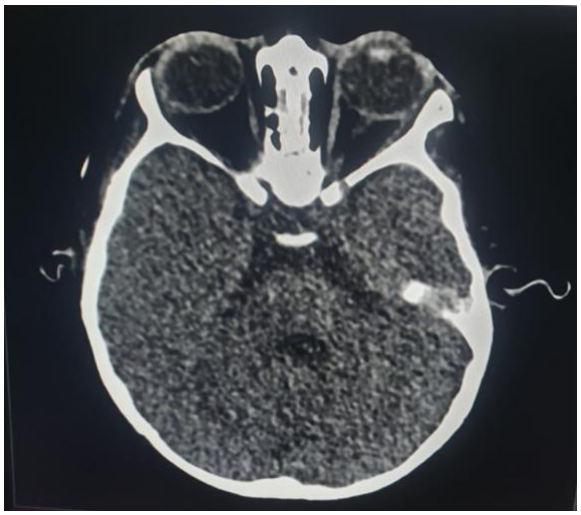

**Fig 3. CT scan with contrast within normal limit**

Follow up examination in 8 months later, There significant progress in visual acuity in order to observation for months. The visual acuity by lea grating is recognized 2.0 CPCM in 60cm that equal 20/300. Retinoscopy showed a cycloplegic refractive error +4.00/-1.25x180 on his right eyes and +4.00/-1.50x180 on his left eyes, and resolve of the horizontal nystagmus. The most notable finding acquired was the patient has shown a delayed development. Despite being 1 year 6 months old age, the patient still crawls and is unable to sit properly**.** Therefore the patient was referred to the division child developmental by pediatrics. By *denver* developmental screening test, they disclosed 4 aspects of delayed developments (social-emotional, language, cognitive and movement) in this child and is later confirmed to have mixed developmental delay. And patient during ophthalmological examination does not show indifference in term of anterior segment**.** There is none provision of intervention treatment in the eye, and monitor the progress of the patient's visual acuity over the next 6 months. In this case were pursue delayed by require larger movement for fine and gross motor skill delay. Training two or three simple words for his speech delay. And evaluation of delay developmental every 6 months to identifying his needs in each developmental area.

## DISCUSSION

Congenitally mydriasis pupil first described by White and Fulton, suggested that it could be due to iris sphincter aplasia. The characteristic of the pupils described as dilated, irregular, and reacted only in the superior temporal and to a lesser extent in the inferior nasal quadrant. [6 7 8]

The etiology remains indefinite, the pathophysiology underlying iris dysgenesis in congenital mydriasis remains unclear and likely reflect many different etiologies. A high

frequency of hypotrophic irides and persistent pupilar membranes are observed in cases by McClelland et al. They described congenital mydriasis associated multisystemic smooth muscle dysfunction syndrome (MSMDS) as mutations of smooth muscle-specific contractile protein alphactin (ACTA2). [10 12 13]

The examined patients from anterior segments show visibly dilated pupils, various scenario of diseases shall be ruled out of this condition. They classified the differential diagnostic as (1). Alteration in ocular structures (isolated congenital mydriasis, hereditary congenital mydriasis, aniridia, Gillespie syndrome, WAGR syndrome). (2). Ocular innervation alteration (bilateral palsy of III cranial oair, parinaud syndrome, congenital horner syndrome, pupila tonic adie). (3). Acquired (traumatic mydriasis infectious mydriasis, pharmacological mydriasis). And in this case we diagnosed to bilateral congenital mydriasis as fixed and dilated pupil at birth without other ocular symptoms with normal vision and without systemic involvement. [9].

In order to know activity of the iris sphincter muscle and see unresponsiveness of the pupils to pilocarpine solutions (0.1% and 2%) eliminated the possibility of central or peripheral pupilloparesis and denervation supersensitivity. However Carrascosa-Romero et al. said most frequent entities in congenital mydriasis differential diagnostic according to new born anisocoria are very infrequent and when appearing are generally more related to ocular innervation alterations than to ocular structure alterations. There is no history of trauma detected on this case, additional examination's result does not suggest a torch and other diseases that lead to conclusion that an organic disease exists. [10 11]

In addition to eyes diseases, delayed development is detected after 6 months of undergoing control visit. Developmental delay indicates extensive deficits and lack of developmental skills that would be appropriate for children at a particular age.[14] However we eliminate organic damage possibility due to negative in torch as most common etiology to developmental delay.[15] Previous research has explained that bilateral congenital mydriasis also suffer from developmental delay. Since an organic disease may have existed as the dilated pupils suggest, we look closely at the pupils through laboratory test, echocardiography, and computed tomography to rule out any coexisting abnormalities, the outcome turns out to be normal. Unavailability of magnetic resonance imaging which hardly to finding neurological deficit in brain and genetic test of ACTA2 to finding exact etiology of this disease. [8 16 17]

Regalado et al. reported there is a relation between congenital mydriasis and delayed development on ACTA2 cases. Twenty-eight congenital mydriasis cases are found among

thirty-three ACTA2 cases. Six delayed development cases are also found as neurologic problem from thirty-one ACTA2 cases. [812]

There a variety of treatments has been given in variant literatures. Brodsky et al. reported 9 years old boy with congenital bilateral mydriasis has visual acuity 20/25 in both eyes. He had no strabismus, nystagmus, or photophobia. He prescribed glasses with +3.00 bifocals fo reading.[18] Rao et al. reports patient who is 24 years old, complaining photophobia, he was prescribed photochromic glasses with bifocal correction.[19] Whilst Caccamise et al. cases 73years old woman realized pupils were always extremely dilated and that this condition had caused her embarrassment during previous ocular examinations when she was questioned about her drug intake. Despite the enlarged pupils the patient has no complaint of photophobia. Her presbyopia symptoms did not appear until she was 45 years old.[7] Other similar case reports presents various complain. Similar cases present various complains. After 8-months of follow up, there is an improved visual acuity, reduced strabismus and horizontal nystagmus. There are none direct symptoms by the patient yet, so we doing observation in 10-months-old as initial assessment to 18-months-old as followed up assessment.

Even the fact that individuals with mydriasis will be extra sensitive to light, with note that the pupils are dilated, wearing the photochromic glasses can be prescribed regardless the photophobic symptoms.[20] We shall evaluate the necessity of giving an addition spectacles to accommodate near sightedness which otherwise cannot be made out of the current constricted pupils.

Based on the cases, it can be said that the prognosis of this disease is varied. Routine cognitive and psychological evaluation should be performed to assess the need for further neurologic evaluation, rehabilitation, and adjustment at school or home. Referring to low vision when regression of visual acquired.[7] Furthermore the patient will follow up every six months to record how far the visual improvement will be.

Many individuals are not aware of their eyes disease due to the lack of visible symptoms and other warnings. Other symptoms found in other parts than the eyes may also be the concerns, for instance the development and growth of the patient also needs to be checked, this raises an urgency for the patients to care with their development with help from pediatrician.

## CONCLUSION

This case bilateral congenital mydriasis accompanied developmental delay without systemic abnormality findings. To be able to diagnose, it should be remembered in the differential diagnosis list of bilateral fixed dilated pupil. Pupillary response test using

pilocarpine solution 0.1% and old photos are helpful in diagnosis. Early detection is significant to screen the need of spectacles to prevent any avoidable vision loss. Comprehensive examination is imperative on the lookout of underdiagnosed diseases which should have gone to therapy.

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