Case Report

# A Case of Congenital Bilateral Anophthalmia

FATEMA ALHUBAISHI[1], AYSHA ALMEDFAA[1], MEHRYAR ANDACHEH[1]

[1]King Hamad American Mission Hospital, A'ali 732, Bahrain

**ABSTRACT:** Introduction. Anophthalmia and microphthalmia are orbito-facial developmental disorders characterized by deficient growth and impaired visual capability [1]. These rare disorders may be unilateral or bilateral. Congenital anophthalmia is the complete absence of the eye [2,3]. The prevalence of both conditions is estimated at 0.2-3 per 10,000 births [4]. We report a case of congenital bilateral anophthalmia that was undetected during follow-up but diagnosed after birth. Case Description. 24-year-old Bahraini female, who is not a known case of any medical illnesses, primigravida at 39+6 weeks of gestation gave birth to a live male baby via vacuum extraction delivery due to recurrent variable decelerations and poor maternal effort. On physical examination, bilateral anophthalmia was immediately observed. No other anomalies were detected. The investigations ordered were MRI brain and orbit, which showed: Absence of bilateral eye globes-features of bilateral anophthalmia. We advised the parents the baby will need socket expansion/ conformer placement to maintain facial symmetry and cosmetic outcome with neurocognitive and development assessment every 2 months as well as speech and language evaluation. Conclusion. Although many probable factors leading to anophthalmia are suggested, many cases arise idiopathically. Due to the nature of the defect, oftentimes prenatal diagnosis with routine scans is challenging. Therefore, more research into probable causes will prompt the healthcare professional to use more sensitive studies to detect the anomaly prenatally to potentially reduce the psychological and financial impact on the parents.

***KEYWORDS:*** *Anophthalmia, Congenital, orbito-facial disorders.*

## Introduction

Anophthalmia and microphthalmia are orbito-facial developmental disorders characterized by deficient growth and impaired visual capability [1].

These rare disorders may be unilateral or bilateral.

They are severe defects that follow a phenotypic spectrum.

Congenital anophthalmia is the complete absence of the eye [2,3].

Microphthalmia is defined as the presence of a hypoplastic or rudimentary eye at birth caused by posterior segment abnormalities like decreased optic cup size or low intraocular pressure.

It may also present as focal ocular malformation like congenital cataract or persistent fetal vasculature [4,5].

The aetiology of these disorders is not yet fully understood.

However, it is hypothesized to be due to deficient formation of the optic vesicle during early weeks of gestation approximately at 6-10 weeks [1].

Anophthalmia and microphthalmia can be isolated or can occur as part of organized syndromes; for example, Fraser, Fryns, Waardenburg or Matthew-Wood syndrome.

Systemic anomalies are detected in at least 50% of individuals [5].

The prevalence of both conditions is estimated at 0.2-3 per 10,000 births [4].

Anophthalmia is further classified into true anophthalmia, in which there is histological absence of ocular tissue in the orbit, and clinical anophthalmia with only tiny or no visible remnants of globe in the orbit [6].

We report a case of congenital bilateral anophthalmia that was undetected during follow up but diagnosed after birth.

## Case Report

We report the case of a 24-year-old Bahraini female, who is not a known case of any medical illnesses, primigravida at 39+6 weeks of gestation (last menstrual period was 10 September 2022) and who presented to our emergency room with labour pains.

She was admitted on 16 June 2023.

Her antenatal follow up was uneventful.

All the relevant blood work was within normal, as seen in Table 1.

A written consent has been obtained from the patient.

A detailed obstetric ultrasound was done on 20 May 2023, which showed a single viable pregnancy corresponding to 36 weeks of gestation, estimated foetal weight was 2.9kg, posterior placenta, Amniotic Fluid Index 14cm, with no gross anomalies detected and a biophysical profile of 8/8.

The patient received two doses of covid vaccine with a third booster dose during pregnancy.





10.12865/CHSJ.50.02.20

***Table 1. Blood investigations.***

| Investigation | Result |
|---|---|
| Blood group | O / Rh - |
| G6PD | Normal |
| Hb | 11g/dl |
| Hb electrophoresis | Normal |
| HBsAg | Negative |
| High vaginal swab | Normal flora |
| Husband blood group | A / Rh - |
| ICT | Negative |
| Maternal test | Result |
| PTC | 186 |
| Rubella | Immune |
| VDRL | Negative |
| WBC | 11.5 |

Note: Blood investigations.

After admission to the labour unit, she progressed well in labour and had a vacuum extraction delivery due to recurrent variable decelerations and poor maternal effort on 16 June 2023 at 01:12PM.

She gave birth to a live male baby in direct occiput anterior position, and the baby cried immediately, weighing 3260g at birth with a length of 50cm.

The third stage of labour was uneventful.

The right medial episiotomy was sutured.

On physical examination, bilateral anophthalmia was immediately detected.

The neonate was active and had a good cry.

Extremities were warm and pink with a good tone and full range of motion.

There were no dysmorphic features. Skin had no rashes and no facial icterus.

Head was normal in size and shape.

Anterior and posterior fontanelles were open.

Ears were normal.

Nose and mouth nares were patent with an intact palate and lip.

Mouth had a well-formed tongue.

Neck and clavicle showed no neck masses and intact clavicles.

Chest was normal in shape and size.

Nipples were normally positioned.

Abdomen and cord were soft, not distended with masses or organomegaly.

The umbilical cord had 2 arteries and 1 vein. There was no umbilical or inguinal hernia.

Hands and feet had 5 digits each with normal creases.

Limbs and hips were stable with good tone and symmetric movement on both lower limbs.

Back showed a straight spine, no sacral dimple and no hair tuft.

Normal external genitalia, anus was patent.

Systemic examination was normal.

## Working diagnosis of bilateral microphthalmia

The investigations ordered were MRI brain and orbit, which showed: Absence of bilateral eye globes-features of bilateral anophthalmia.

Tiny fluid signal intensity was noted in the right side along the anterior aspect of bony orbit which may represent rudimentary eyeball.

Bony orbital walls were normal with presence of extraocular muscles.

Mildly dilated 4th ventricle-connected to cisterna magna suggesting a possibility of dandy walker variant.

Another differential diagnosis was blakes cyst.

However, there was no evidence of hydrocephalus, no significant hypoplasia of vermis or cerebellar hemispheres.

Corpus callosum-genu, body and splenium appear normal.

Rostrum of corpus callosum was also visualized, though poorly characterized.

Questionable ectopic posterior pituitary location-hypersensitivity seen in higher position. Pituitary stalk interruption syndrome cannot be excluded.

The MRI scans are represented in Figure 1.

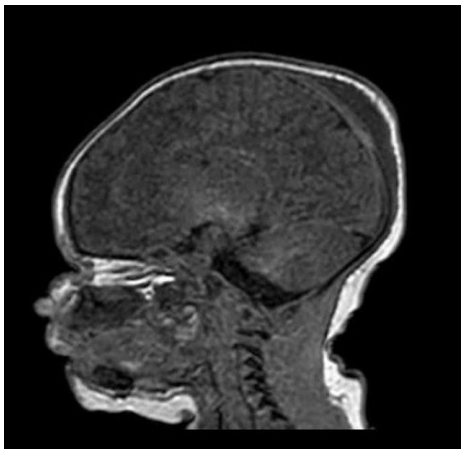

***Figure 1. Sagittal MRI cut of the brain showing posterior displacement of the globes bilaterally.***

Ultrasound abdomen was normal except for dilated left renal pelvic with collapsed proximal ureter, diameter around 7-8mm.

Ultrasound scans are displayed in Figure 2.

She was tested for auditory brainstem response and was normal bilaterally.

We advised the parents the baby will need socket expansion/conformer placement to maintain facial symmetry and cosmetic outcome with neurocognitive and development assessment every 2 months as well as speech and language evaluation.

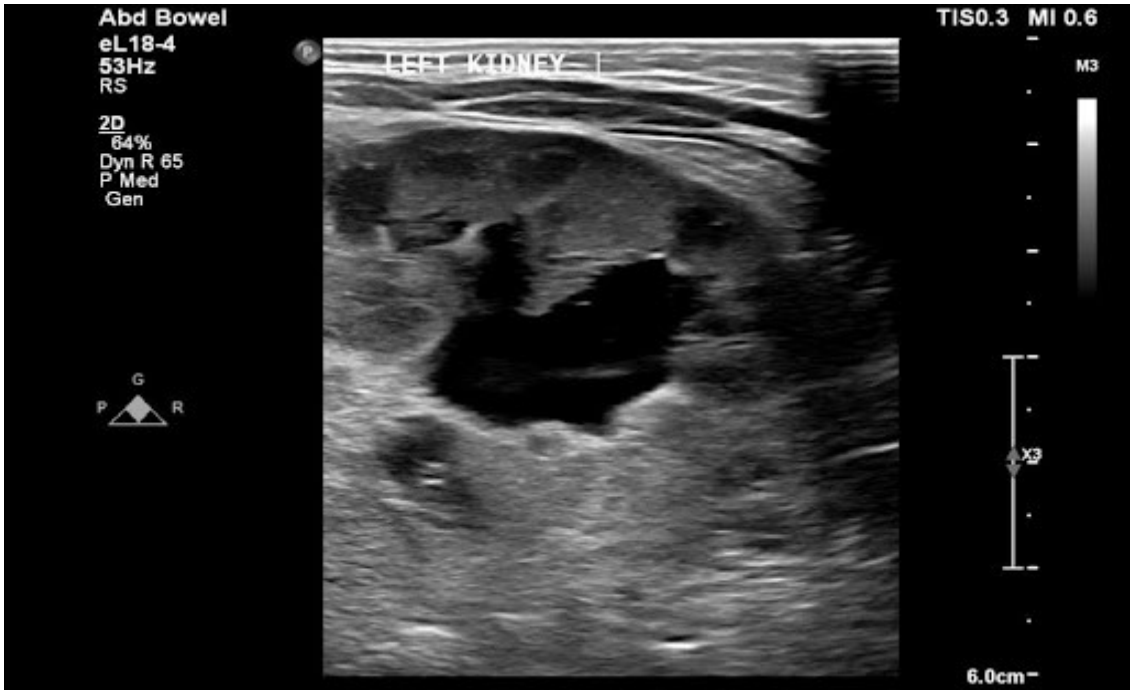


***Figure 2. U/S abdomen showing dilated left renal pelvis.***

## Discussion

Multiple maternal and gestational variables, genetic alterations and environmental factors have been studied for possible risk factors but none were found [6].

Absence of globes causes orbital hypoplasia as well as hemifacial microsomia; mainly affecting the maxilla and the mandible.

Bilateral cases appear with sunken orbits and midfacial hypoplasia [6,7].

Anophthalmia and microphthalmia are diagnosed clinically by ophthalmological examination.

Once a diagnosis is made both ocular and systemic imaging modalities must be done including Ultrasound, CT and MRI to rule out other systemic abnormalities such as neurological, renal, cardiac or others.

Treatment is dependent on aetiology and severity [8].

Mild cases are followed up regularly for orbitofacial growth [1,9,10].

Patients with severe microphthalmia or anophthalmia, without potential vision, need early orbital rehabilitation with "artificial" stimulation of orbitofacial growth [11].

Our patient needed further investigations and was offered management options.

However, her parents opted to seek medical care in another facility due to financial reasons.

Although many probable factors leading to anophthalmia are suggested, many cases arise idiopathically.

Due to the nature of the defect, oftentimes prenatal diagnosis with routine scans is challenging.

Therefore, more research into probable causes will prompt the healthcare professional to use more sensitive studies to detect the anomaly prenatally to potentially reduce the psychological and financial impact on the parents.

### Acknowledgments

We would like to thank Dr. Zainab Yusuf, Specialist Radiologist certified in diagnostic radiology for her contribution.

### Conflict of interests

None to declare

### References

1. Verma AS, FitzPatrick DR. Anophthalmia and Microphthalmia. Orphanet J Rare Dis, 2007, 2:47.
2. Tucker S, Jones B, Collin R. Systemic anomalies in 77 patients with congenital anophthalmos or microphthalmos. Eye (Lond), 1996, 10(Pt 3):310-314.
3. Bardakjian TM, Schneider A. The genetics of anophthalmia and microphthalmia. Curr Opin Ophthalmol, 2011, 22(5):309-313.
4. Slavotinek AM. Eye development genes and known syndromes. Mol Genet Metab, 2011, 104(4):448-456.
5. Shaham O, Menuchin Y, Farhy C, Ashery-Padan R. PAX6: A multi-level regulator of ocular development. Prog Retin Eye Res, 2012, 31(5):351-376.

 10.12865/CHSJ.50.02.20

6. Hayashi N, Repka MX, Ueno H, Iliff NT, Green WR. Congenital cystic eye: report of two cases and review of the literature. Surv Ophthalmol, 1999, 44(2):173-179.
7. Amrith S, Lee YS, Lee JJ, Liew GC, Leo SW, Khoo BK. Congenital orbito-palpebral cyst in a case of Fraser syndrome. Orbit, 2003, 22(4):279-283.
8. Golzio C, Martinovic-Bouriel, J, Thomas S, Mougou-Zrelli S, Grattagliano-Bessieres B, Bonniere M, Delahaye S, Munnich A, Encha-Razavi F, Lyonnet S, Vekemans M, Attie-Bitach T, Etchevers HC. Matthew-Wood syndrome is caused by truncating mutations in the retinol-binding protein receptor gene stra6. Am J Hum Genet, 2007, 80(6):1179-1187.
9. Traboulsi EI. Developmental genes and ocular malformation syndromes. Am J Ophthalmol, 1993, 115(1):105-107.
10. Boyd PA, Armstrong B, Dolk H, Botting B, Pattenden S, Abramsky L, Rankin J, Vrijheid M, Wellesley D. Congenital anomaly surveillance in England-ascertainment deficiencies in the national system. BMJ, 2005, 330(7481):27.
11. Morrison D, Fitzpatrick D, Hanson I, Williamson K, van Heyningen V, Fleck B, Jones I, Chalmers J, Campbell H. National Study of Microphthalmia, Anophthalmia, and Coloboma (MAC) in Scotland: Investigation of Genetic Aetiology. J Med Genet, 2002, 39(1):16-22.

***Corresponding Author: Fatema Alhubaishi, King Hamad American Mission Hospital, A'ali 732, Bahrain***
***e-mail: fatemaalhubaishi@gmail.com, fatema.alhubaishi@amh.org.bh***