Rare Case of Idiopathic Congenital Unilateral Anophthalmia

Dr Murtuza Jhabuawala¹, Dr. A.P.Agashe², Dr. Mehul Vikani³, Dr. Sagar Shetty⁴, Dr. Mikeen Shah⁵

ABSTRACT

True or primary anophthalmos is very rare. Anophthamia whether acquired or congenital, is not just a cosmetical worry. Absence or loss of eye can affect maturation of surrounding soft tissues and bony structures of the affected orbit as well loss of stereopsis if unilateral. A case of idiopathic congenital unilateral anophthalmia is been reported. A 26 year old male came to us at our hospital with absence of left eyeball since birth.

Keywords: Anophthalmia, Congenital, Idiopathic, Unilateral

¹Resident, ²Professor, Dept of Ophthalmology ³Resident, Dept. of General surgery, ⁴Department of radio diagnosis, MGM Hospital, Navi Mumbai, India

⁵Resident, Aravind Eye Hospital, Pondicherry, India

Corresponding author mail: <u>murtu.95@gmail.com</u>

Source of support: Nil Conflict of interest: None declared

INTRODUCTION

True or primary anophthalmos is very rare. Only when there is a complete absence of the ocular tissue within the orbit can a diagnosis of true anophthalmos be made. Anophthalmia occurs when the neuroectoderm of the primary optic vesicle fails to develop properly from the anterior neural plate of the neural tube during embryological development. Proper growth

of the orbital region is dependent on the presence of an eye, which stimulates growth of the orbit and proper formation of the lids and the ocular fornices. Commonly, a child born with anophthalmia has a small orbit with narrow palpebral fissure and shrunken fornices.

CASE REPORT: A 26 year old male presented to us with absent left eyeball. Patient gave history of having the same

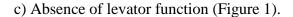
condition since childhood. He gave no history of trauma or any positive family history.

On examination:

1. Globe findings: Left globe was completely absent (Figure 1).

2. Orbital findings:

- a) Small oribital rim and small size of the orbital cavity was noted (Figure 3).
- b) Extra ocular muscles were absent (Figure 3).



3. Eyelid findings:

- a) Very small palpebral fissure (Figure 1).
- b) Shallow inferior conjunctival fornix (Figure 2).
- c) Shortening of the upper and lower eyelids.

4. Systemic examination:

No abnormalities were detected systemically.





Figure 1 Figure 2



FIGURE 3

INVESTIGATIONS

An USG B Scan was done which confirmed our findings of absence of left globe (figure 4).

MRI and CT Scans could not be taken.



Figure 4

DISCUSSION

Anopthamia refers to absence of ocular tissue in the orbit. Little is known about the etiology of anophthalmia.

From the clinical studies of various reported cases these facts were noted. Anophthalmia in man is one of the rare congenital abnormalities. Anophthalmia has been reported to be present in 3 out of every 100,000 births. Congenital anophthalmos is a very rare condition that has a reported

prevalence rate of 0.18 per 10,000 births in the United States, 0.19 case per 10,000 births in European countries, 06-0.42 case per 10,000 births in Australia. (1,2,3,4)

Anophthalmia can be unilateral or bilateral. Causes of anophthalmia include idiopathic, genetic, chromosomal and prenatal environment.

It can be classified as follows

Primary where there complete absence of the eyeball to form due to failure of the brain to form the tissue.

Secondary/degenerative in which the development starts initially and then stops leaving the child with only a small amount of residual ocular tissue.

SUMMARY

Rare case of "Idiopathic Congenital Unilateral Anophthalmia" is been reported here. It can be a very debilitating condition for the child. Still the etiology of Anophthalmia is uncertain, further studies need to be conducted to confirm the etiology.

ACKNOWLEDGEMENT

We would like to extend our heartfelt gratitude to the Department of Ophthalmology and Department of Radiology.

REFERENCES

- 1. Verma, Amit S; Fitzpatrick, David R (2007). "Anophthalmia and microphthalmia". Orphanet Journal of Rare Diseases 2: 47. doi:10.1186/1750-1172-2-47.
- 2. Shaw GM, Carmichael SL, Yang W, Harris JA, Finnell RH, Lammer EJ. Epidemiologic characteristics of anophthalmia and bilateral microphthalmia among 2.5 million births in California, 1989-1997. Am J Med Genet A. Aug 15 2005; 137(1): 36-40.
- 3. Boyd PA, Haeusler M, Barisic I. EUROCAT Report 9: Surveillance of congenital anomalies in Europe 1980-2008. Birth Defects Res A Clin Mol Teratol. Mar 2011; 91 Suppl 1:S1.
- 4. Skalicky SE, White AJ, Grigg JR, et al. Microphthalmia, anophthalmia, and coloboma and associated ocular and systemic features: understanding the spectrum. JAMA Ophthalmol. Dec 2013; 131(12): 1517-24.