Case Report

Two Cases of Anophthalmia and Quality Of Life

Abstract:
The absence of one or both eyes was called anophthalmia included the globe eye and the ocular tissue being missing from the orbit. Anophthalmia is an extremely rare disease and is rooted in genetic abnormalities. It can also be associated with other syndromes. This paper presents 2 cases with anophthalmia. One is no treatment. The other with some treatments are discussed contributing the diversifying clinical signs. Both cases with patient's quality of life are noted in developing country.
Key words: Anopthalmia, microphthalmia, other disorders, quality of life

1. Introduction:

Anophthalmia is the absence of one or both eyes. Both the globe (human eye) and the ocular tissue are missing from the orbit. The absence of the eye will cause a small bony orbit, a constricted mucosal socket, short eyelids, reduced palpebral fissure and malar prominence. [1]. With 2018 ICD-10-CM, diagnosis code Q11.1 Problems during embryonic development can lead to congenital eye malformations, such as anophthalmia (no eye), microphthalmia (small eye), coloboma (failure of the optic fissure to close), aniridia (absent or partial iris), and optic nerve hypoplasia (underdeveloped optic nerve) is for anophthalmos, microphthalmos and macrophthalmos. Genetic mutations, chromosomal abnormalities, and prenatal environment can all cause anophthalmia. Anophthalmia is an extremely rare disease and is rooted in genetic abnormalities. It can also be associated with other syndromes.

Epidemiology: Anophthalmia has been reported to be present in 3 out of every 100,000 births [2]. A recent study in the UK indicated that anophthalmia and microphthalmia had a combined average of 1 in every 10,000 births [3]. The annual rate of occurrence of anophthalmia/microphthalmia in the United States is about 780 children born/year [3]. Parents that already have a child who suffers from anophthalmia has a 1 in 8 chance of having another child with anophthalmia [4]. Approximately 2/3 of all cases of anophthalmia are determined to be of genetic basis. Causes: SOX2: The most genetic based cause for anophthalmia is caused by the SOX2 gene. Sox2 anophthalmia syndrome is an autosomal dominant inheritance. There are at least 33 mutations in the Sox2 gene that have been known to cause anophthalmia [5]. RBP4: has recently been linked to autosomal dominant form of anophthalmia. [6]

Comment [A1]: Uniform the writing of "SOX2".

Other influential genes: SOX2 and RBP4 are not the only genes that can cause anophthalmia. Other important genes include OTX2, CHX10 and RAX. Each of these genes is an important in retinal expression. OTX2 is dominantly inherited. Environmental influence: Many environmental conditions have also been known to cause exophthalmia such as children with gestational-acquired viral infections, Toxoplasma, rubella, and certain strains of the influenza virus. Besides environmental conditions tat have led to anophthalmia are maternal vitamin A deficiency, exposure to X-rays during gestation, solvent abuse, and exposure to thalidomide. [3] Chromosome 14: An interstitial deletion of chromosome 14 has been known to occasionally be the source of anophthalmia. [7]

Anophthalmia is one of the leading causes of congenital blindness and accounts for 3-11% of blindness in children [8]. Anophthalmia and microphthalmia together make up 1.7-1.8% of reconstructive surgical cases of plastic surgery and ocular prostheses. [9]

This paper presents 2 cases with anophthalmia. One is anophthalmia /microphthalmia with some treatments are discussed contributing the diversifying clinical signs. Other is anophthalmia /microphthalmia on a disabled child and he was not anytreatment. Both cases with patient's quality of life are noted in developing country.

2. Case Report: (Fig 1 & 2. attach)

Case 1: Anophthalmia associated with other disorders. . An 7 year old man with an anophthalmia associated with other disorders such as mental less development, malformation of nose, lip, abnormal

Comment [A2]: Please correct the punctuation.

hand and leg (Figure 2) is presented. He is a severe disorders in mental less development. He is always laying on bed. General examination:
General state: Height= 1 meter; Weight=20 kg; Pulse = 75/mn; Arterial tension=120/75 mmHg. Mental =less development. He can not speak, can not move. Ophthalmology examination:
Right eye (RE) = no eye. Left eye (LE) = small eye with abnormal ocular layers from cornea to sclera, choroid and retina. Both eyes (OU) =Blindness.
Diagnosis: RE=Anopthalmia and LE=microanophthalmia associated with other disorders
Treatment: The treatment is not performed
Follow up: Quality of life is not good
He is living in the house for orphans and disabled children of Buddhist Temple –conducted by a non-government organization. There are many "Love Houses" like this in many provinces and cities in Vietnam,.
Case 2: Anophthalmia/microphthalmia An 18 years old man with an anophthalmia is presented. He is a pupil at a special high school for the blind in HCM city (Founded 1926) . He is checked up his health period 6 months/ a time.
General state: Height= 1, 50 meter; Weight=50 kg; Pulse = 75/mn; Arterial tension=120/75 mmHg.
Mental = normal development. Nothing abnormal detecting (N/A)
Ophthalmology examination:
Right eye (RE) = no eye. Left eye (LE) = small eye with abnormal ocular layers from cornea to sclera, choroid and retina. Both eyes (OU) =Blindness.
Diagnosis: RE=Anopthalmia and LE=microanophthalmia
Treatment: Surgery RE for artificial eye for the esthetic view
Follow up: Quality of life is still good.

Discussion:

There are three classifications for anophthalmia:

• Primary anophthalmia is a complete absence of eye tissue due to a failure of the part of the brain that forms the eye.
• Secondary anophthalmia the eye starts to develop and for some reason stops, leaving the infant with only residual eye tissue or extremely small eyes which can only be seen under close examination.
• Degenerative anophthalmia the eye started to form and, for some reason, degenerated. One reason for this occurring could be a lack of blood supply to the eye

Prenatal diagnosis:
Ultrasounds can be used to diagnose anophthalmia during approximately 20 weeks gestation. With 3D and 4D ultrasounds have proven to be more accurate at viewing the fetus's eyes during pregnancy.
Amniocentesis can only diagnose anophthalmia when there is a chromosomal abnormality. Chromosomal abnormalities are only a minority of cases of anophthalmia.
Postnatal diagnosis
MRIs and CTs can be used to scan the brain and orbits included the internal structures of the globe, the optic nerve and extraocular muscles, and brain anatomy.
Examination: Aside from these associative conditions that are: Lenz Syndrome, Goldenhar-Gorlin Syndrome, Waardenburg syndrome, anophthalmia in only one eye tends to be associated with complications in the other eye. These risks include a higher chance of having glaucoma or a detached retina.
Treatments: Currently, there is not a treatment option for regaining vision by developing a new eye. The child will need to go to a prosthetic eye for cosmetic options. Cosmetic surgery: If the deformities do appear, the surgery is not done until at least the first two years of life. Many people get eye surgery, such as upper eyelid ptosis surgery and lower eyelid tightening. These surgeries can restore the function of the surrounding structures like the eyelid in order to create the best appearance possible [9].
In the 1st case: anophthalmia associated with other disorders and he did not have any medical intervention. He stays always on bed with helping all activities by voluntees in Buddhist Temple.

Comment [A3]: The classifications for anophthalmia needs reference.
I suggest including references to "Ultrassounds", "Amniocentesis" and "Associative Conditions" too.
At the end of the discussion, the author could mention the need for health professionals to be prepared to deal with anophthalmia, as can be seen in the abstract of this article, which is in English.
https://www.scielo.br/j/rbof/a/nxCHp6dbWjHW6cxvTTMpMSb/?lang=pt

Comment [A4]: This comment needs an reference.

The 2nd case is 18 year old, blindness totally with mental well development. Right eye (RE) = no eye. (Primary anophthalmia); Left eye (LE) = Microphthalmia with abnormal ocular layers from cornea to sclera, choroid and retina. He was prepared for prosthetic in left eye in order to have got both eyes in esthetic view. This patient is a blind person. But he is learning, is participating all communicable activities with the same age people by helping of family as well as society. In Vietnam, there are many blind schools in many provinces and cities. This is a reason make our patient decreasing worry about his disability. (Fig 3 attach)

Quality of life: The period of follow-up of 1st patient was over 6 years. He was a severe disabled child and he has been nourished in the "Love House" of Buddhist Temple. His quality of life is not good but with the take care by community can help his family a lot of fee as well as the love of everybody in Buddhist Temple. The period of follow-up of 2nd[t] patient post intervention in this paper was 10 years. He is continuing to learn, to play, and to participate all activities with the same age in a special school for blind people All of these as a part of palliative care, can be a reason make the patients decreasing worry about their disability. Despite of a developing country the quality of life index of Vietnam is ranged 60 in 2018. Numbeo is the world's largest database, provides current and timely information on world living conditions including cost of living, housing indicators, health care, traffic, crime and pollution. [10].

Conclusion:

In the 1st[nd] case his quality of life is not good but with the take care by community can help his family a lot of fee as well as the love of everybody in Buddhist Temple.

This is a reason make the patient decreasing worry about his/her disability contributed better quality of life for patients as well as family.

This 2nd case is 18 year old, blindness totally contributing of diversifying clinical eye signs but mental well development as well as physical nearly normal development. So prosthetic in his left eye to have got both eyes in esthetic view can make a better quality of life. The patient has to learn, to participate all activities with the same people age.

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Conflict of Interest: No financial disclosures

References

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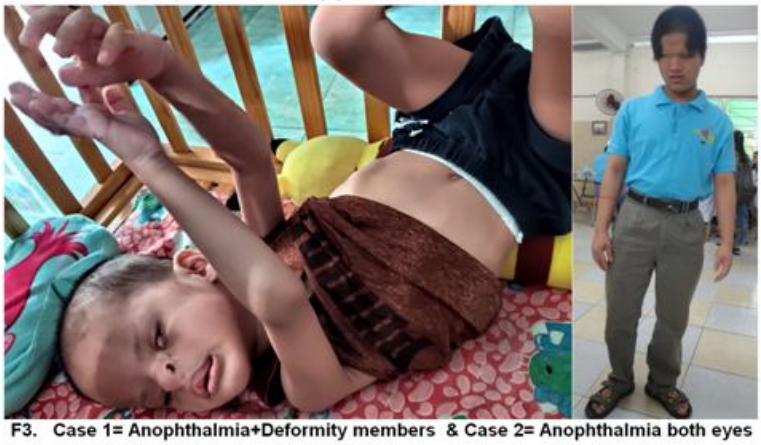
Comment [A5]: The inclusion of the suggested reference in the discussion will contribute to broaden the message of the scientific work.

Comment [A6]: Please complete this reference.

F1. Case1= Anophthalmia + Ocular Atrophy + Other disorders

F2. Case 2 with artificial eye



F3. Case 1= Anophthalmia+Deformity members & Case 2= Anophthalmia both eyes

Comment [A7]: I did not find the figures mentioned in the text. Please review this.

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