Case Report: Anophthalmia

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Abstract:
Anophthalmia is the absence of one or both eyes. Both the globe (human eye) and the ocular tissue are 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 a case with anophthalmia /micropthalmia with some treatments are discussed contributing the diversifying clinical signs and patient's quality of life.
Key words: *Anopthalmia, microphthalmia, quality of life.*

1. Introduction:

Anophthalmia is the absence of one or both eyes. There are missing from the orbit, the ocular tissue. The causes included a small bony orbit, a constricted mucosal socket, short eyelids, reduced palpebral fissure and malar prominence in the absence of the eye With 2018 ICD-10-CM, diagnosis code Q11.1 we have: 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. during embryonic development. Anophthalmia causes consist of genetic mutations, chromosomal abnormalities, and prenatal environment 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 have 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 the autosomal dominant form of anophthalmia. [6]
Other influential genes: SOX2 and RBP4 are not the only genes that can cause anophthalmia. Other important genes include OTX2, CHX10 and RAX. OTX2 is dominant inheritance. These genes are an important retinal expression
Influence from the environment: The exophthalmia children with gestational-acquired viral infections, toxoplasma, rubella, and certain strains of the influenza virus may be caused by environmental conditions
Besides environmental conditions, anophthalmia can be caused by maternal vitamin A deficiency, exposure to X-rays during gestation, solvent abuse, and exposure to thalidomide. [3]
Chromosome 14: The source of anophthalmia has occasionally been known by an interstitial deletion of chromosome 14. [7]
There are 3-11% of congenital blindness in children in anophthalmia [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 a case with anophthalmia/microphthalmia with some treatments are discussed contributing to the diversifying clinical signs and patient's quality of life

2. Case Report: (Fig. attach)

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.

3. Discussion:

There are three classifications for anophthalmia:
Primary anophthalmia with complete absence of eye tissue caused by a failure of the part of the brain that forms the eye.
• Secondary anophthalmia: the eye develops and after that stops for some reason. The infant with only residual eye tissue or extremely small eyes can be seen under examination.
• Degenerative anophthalmia: the eye began to develop and, after that for some reason, degenerated. This occurring could be a lack of blood supply to the eye.
Prenatal diagnosis:
With 3D and 4D ultrasounds have been done to be more precise at viewing the fetus's eyes during pregnancy. Ultrasounds diagnosis of anophthalmia is performed during approximately 20 weeks gestation.
Amniocentesis can only diagnose anophthalmia when there is a chromosomal abnormality. Only a minority of cases of anophthalmia.is chromosomal abnormalities.
Postnatal diagnosis:
MRIs and CTs diagnosis can be done to scan the brain and orbits included the internal structures of the globe, the optic nerve and extraocular muscles, and brain anatomy. In clinical sigins anophthalmia with lenz syndrome, Goldenhar syndrome, Waardenburg syndrome,. in one eye to be often associated with complications in the other eye. Besides, glaucoma or a retinal detachment are risks higher chance in association
Treatments: Currently, there is no treatment option for vision. So cosmetic options for the child will need to choose. Cosmetic surgery: If the deformities do appear, the surgery should be done at the first two years of the life of the child. The surgery included: The upper eyelid ptosis, the lower eyelid tightening and orbital repairing. These surgeries can restore the function of the eyes as well as create the best appearance possible [9].
This case is 18 years old, blindness totally with mental good development. Right eye (RE) = no eye, (Primary anophthalmia); Left eye (LE) = Microphthalmia with abnormal cornea to sclera, choroid and retina. He was prepared for a prosthetic in his left eye to have got both artificial eyes in appearance view.
This patient is a blind person. But he is learning, is participating in all communicable activities with the same age people by helping of a family as well as society. In Vietnam, there are many blind schools in many provinces and cities. This is a reason to make our patient decrease worry about his/her disability. The quality of life index of Vietnam is ranged 60 in 2018, in developing countries. Numbeo is the world's largest database with the conditions including cost of living, housing indicators, health care, traffic, crime and pollution were timely provided. [10]

Conclusion: This case is 18 years old, total blindness contributing to diversifying clinical eye signs but mental good 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 in all activities with the same people age. This is a reason to make the patient decrease worry about his/her disability contributed better quality of life.

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RE= Anophthalmia-LE= Microanophthalmia

Author Profile

Duong Dieu received the MD (1978) and PhD (2003). He was chief of the Ophthalmology Department for over 30 years with a clinician/surgeon. From 2010 to now he is vice dean of the Faculty of Medicine of Nguyen Tat Thanh University in Hochiminh city- Vietnam.