Introduction

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/microphthalmia with some treatments are discussed contributing the diversifying clinical signs and patient’s quality of life.

Keywords: anophthalmia · microphthalmia · quality of life

Epidemiology

Anophthalmia has been reported to be present in 3 out of every 1,00,000 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

It has recently been linked to 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. 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 that 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 a case with anophthalmia/ microphthalmia with some treatments are discussed contributing the diversifying clinical signs and patient’s quality of life (Figure 1).

Case Report

An 18-year-old man with an anophthalmia is presented. He is a pupil at high blind school. 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 Development : normal. Nothing abnormal detecting (N/A).

Diagnosis

RE: Anophthalmia 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 no 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].

This 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.

Figure 1. RE - Anophthalmia, LE - Microanophthalmia

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.
This patient is a blind person. But he is learning, participating all communicable activities with the same age people by helping of family as well as society. This is a reason make our patient decreasing worry about his/her 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

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

### References