

## Keep an Eye on Fetal Anophthalmia Detection

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### Incidence

Anophthalmia represents a rare entity that affects 3/1000 live born infants and it accounts for 4 - 5% of congenital blindness. In such cases, the eye globe of the fetus is not present due to the failure of the optic pit to create a vesicle (primary) or when the optic vesicle forms and degenerates (secondary anophthalmia) [1].

### Etiopathogenesis

Even though most lesions are bilateral, it may occur as a unilateral defect. The disease appears to the same degree in both sexes and it is mostly associated with central nervous system (CNS) malformations, trisomy 13, cytomegalovirus and toxoplasmosis infection or a genetic syndrome [1].

Although, the exact etiology is vague, the increased maternal age has been implicated.

Goldenhar syndrome is characterized by the coexistence of anophthalmia, fascial clefts, ear lesions and underdeveloped internal organs. Moreover Fraser syndrome represents one of the commonest autosomal recessive conditions in which anophthalmia coexists with microphthalmia, cardiac defects, bilateral renal agenesis, trachea atresia, fascial clefts and syndactyly.

The prognosis depends on whether anophthalmia is an isolated defect or it is associated with other malformations (> 50% of cases) [1,2].

### 2D, 3D, 4D ultrasonography or MRI

High resolution transvaginal sonography has been a major diagnostic tool in the prenatal detection of lesions of orbits and lens at 12 weeks of the gestation.

Campbell, *et al.* firstly described the 3D image (reverse face) to evaluate fascial cleft palate [3]. Interestingly, Wong, *et al.* used this technique to detect anophthalmia in cases where fetal head was not in the correct position and plane [4].

According to the current literature, anophthalmia is traditionally diagnosed by two-dimension (2D) ultrasonography in the first or second trimester during pregnancy. The orbits and the lens of the fetal eyes can be viewed in the coronal planes when the head is in an occiput posterior position.

In several cases, the absence of one or both eyes is associated with microphthalmia, hypotelorism or hypertelorism. Physicians should measure the orbital diameter (10 mm), the intraocular and binocular distance of the fetus guided by the 2D scan, in order to exclude hypoplastic or rudimentary eyes at the anomaly scan. In normal circumstances, as a rule of thumb, the interorbital distance is equal to each ocular diameter.

Fetal magnetic resonance (MRI) is preferred in order to confirm the absence of the eye tissue, the extraocular muscles and optic nerve. Additionally, in the third trimester MRI can further detect CNS malformation such as agenesis of sulci and gyri.

Although, 3D and 4D ultrasonography have proven to be more accurate compared to 2D scan (avoiding shadowing effects), it should be preferred as adjuvant technique to confirm the defect [2,5].

### Management

To conclude, if anophthalmia coexists with cardiac, CNS and musculoskeletal defects, clinicians should consider termination of the pregnancy.

Otherwise, in isolated forms, Ophthalmologists should perform ocular prosthesis in order to avoid deformities during growth and preserve normal development of bones.

Thus, a prenatal detection of an absent eye is crucial regarding the prognosis and neonatal management.

### Bibliography

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