

**ALOBAR HOLOPROSENCEPHALY IN
1ST TRIMESTER PREGNANCY**



Table of Contents

Background..... 3

Signs and Symptoms Of Holoprosencephaly 3

Case Description 5

Ultrasound Findings..... 6

Conclusion 7

References:..... 8



Background

Ionescu et al. (2018) described Holoprosencephaly as a malformation in the brain's structure, which results from the incomplete union and development of the two hemispheres of the brain. An article published by Medline Plus (2022) adds that it is a condition that affects the development of the head and face. Holoprosencephaly is abnormal in brain development because the brain is not properly divided into left and right hemispheres. The condition can also affect the development of the head and face. There are 4 types of Holoprosencephaly, which are distinguished by their severity. From the most severe to the smallest, there are 4 types of leaf length changes, leaf variability, leaf length changes, blisters, and length changes between them (MIHV) (Kruszka and Muenke, 2018).

The severity of any abnormality on the face generally corresponds to the severity of the abnormal brain. Those severely damaged have a central eye (one eye) and a tubular nasal structure (knot) above the eye. The brain is only partially isolated in its mildest form, and the eyes are usually close together. Other signs and symptoms are often developmental and pituitary problems (Dubourg et al., 2018). Mutations in at least one of the 14 different genes can cause brain damage; chromosomal defects; or medicines that can cause birth defects (malformations). It can also be a symptom of a single genetic syndrome. In many cases, the exact cause is unknown. Life expectancy for people with this disorder varies, and treatment depends on each person's symptoms and severity.

Signs and Symptoms Of Holoprosencephaly

Depending on the severity of the malformation, holoprosencephaly can be divided into three main subtypes: lobar, hemilobaric and lobar, and there is a fourth subtype called

interhemispheric variation (MIH): Holobrocephaly Alobar is the complete inability of the brain to divide the left and right hemispheres of the brain, leading to the loss of the midline structures of the brain and face and the joining of the chambers of the brain called the lateral and third ventricles. Ventricles (usually separately) (Roessler, Hu and Muenke, 2018). Manifestations of the face may be one eye (cycloplegia) or closed eyes (chest cavity) or no eyes (anophthalmia) or very small eyes (microphthalmia), a tubular nose (beak); or eyes too far between a closed nose (low) and a flat or split lip in the middle of the lip (middle lip gap) or on both sides (bilateral lip gap). In some cases, the face looks almost normal, especially in people with a variant (mutation) of the ZIC2 gene.

An anterior cerebral membrane failure occurs when the left side of the brain merges with the right part of the brain, called the anterior (anterior) and lateral leaves (both sides of the brain) (Monteagudo, 2020). There is also a separation between the left and right hemispheres of the brain (known as the retinal gap) just behind the brain. Patients with Holoprosencephaly may have myopia, microphthalmia or anophthalmia. Other features may include a flat bridge and forehead, nasal, medium or bilateral cleft lip and palate.

Holoprosencephaly lobar refers to two ventricles (left and right), but the hemispheres are connected to the anterior cortex (Kim et al., 2019). Signs may include bilateral lips, eyes closed, a sunken nose, or an almost normal face. The variant in the hemisphere's middle occurs when the brain joins in the middle. Symptoms may include closed eyes, dizziness, a narrow nose, or an almost normal appearance. Other signs and symptoms may include seizures, hydrocephalus, neurological defects, pituitary dysfunction, short stature, difficulty eating, unstable body temperature, heart rate, and breathing (Kruszka and Muenke, 2018). Most patients with Holoprosencephaly have developmental and developmental disorders, which depend on the

severity of the brain problem. Alobar holoprocentphalia is a subtype of holoprocentephalus and is the most severe of the three classic subspecies, with milder clinical signs of semi- and lobar-holoprosencephaly.

Case Description

The case deals with some major aspects of the Alobar Holoprosencephaly in 1st Trimester of Pregnancy. The condition seems a bit challenging and confusing due to the widespread issues. As mentioned above, the issue of Alobar Holoprosencephaly in 1st Trimester of Pregnancy is a complicated matter to handle. This leads to increased chances of premature death. Therefore, the case revolved around some major aspects that were considered special.

The identification of the case came into being after considering the Patient's regular checkup routine. The Patient was booked for the routine nuchal. As per the normal; time, the scanning was done for the lady. In the stars, everything was okay, but later in ultrasound, some major aspects were found about the child. There was an identification of the Alobar

Holoprosencephaly in 1st Trimester of Pregnancy. After consideration, the overall inspection took the place of the situation.

It was also found from the Patient's history that before the scanning for the ultrasound, patients were facing insulin-dependent that is of type 1 diabetes. Concerning pregnancy, type 01 diabetes is considered one of the major and most complex cases to handle. This has a direct impact on the child's growth and development. As a result, the patients had some major and essential aspects to consider. For instance, the prior scan occurs due to poor insulin control at conception. In such a manner, the situation became more complicated. Although from the demographic profile of the lady, it was clear that the Patient was a 34 years old black Caribbean

lady. She gained some experience from her previous pregnancy as she suffered 3 pregnancies before this one having 3 live births and the children. The premature situation was also one of the major challenges to handle. The Patient was with 13 weeks and 3 days for the pregnancy, which is quite a complicated time under such conditions.

This is how the overall case was presented to the healthcare professionals. The case clearly shows various aspects to consider, such as Alobar Holoprosencephaly in 1st Trimester pregnancy, the age of the lady, her diabetic disorder and other aspects. All of them gained great attention from the professionals. The ultrasound also provided some great information about the situation that needed to be handled. Therefore, the thesis was claimed as one of the most complicated cases to handle under such circumstances.

Ultrasound Findings

Concerning the ultrasound report, numerous factors got clear under the circumstances. Therefore, proper scanning has taken place with visible outcomes. During the scan, it was visible that the foetus was growing as expected, with the Crown Rump Length (CRL) measuring 74.2mm which approximately dates the pregnancy as 13 weeks +3 days. This showed that the pregnancy was growing as expected and correlated with the Patient's dates. On the other hand, further findings in this regard also revealed that there were some observations during the ultrasound as per local protocols. This observation has claimed that the heart was visualised and beating normally. This was one of the most satisfactory results.

In addition, the stomach was visible. As part of this, both the hands and feet of a child were visualised. Moreover, the Abdominal wall appeared normal in the ultrasound. Apart from this, there was an unexpected find while scanning for the head to visualise the brain. In such a

case, the normal physique of the brain was not visualised. Initially thought to be positional, the sonographer changed position before trying again. Still, the image was similar with such specific changes. The patient was referred for a 12- week screening scan based on that. The scan has shown unexpected development in the foetus of suspicious of an early sign of alobar Holoprosencephaly. A second sonographer confirmed these findings.

The case that has been discussed in this regard has some complicated and technical requirements regarding child development. However, the situation seems supportive and clear. There was no specific measure that could impact the overall finding of the results. Still, the proper inspection was made that provided a clear understanding of the situation. It was also observed and learned that the guideline protocols are considered essential and specific measures to be followed. The findings were made in the ultrasound, but as per department protocol, any abnormalities or anomalies seen must be confirmed by a second sonographer. Therefore, proper inspection has confirmed the issues while using a second sonographer.

Conclusion

In this report, a case has been discussed with Holobrocephaly Alobar for 13 months and 3 days of pregnancy. Under the condition of ultrasound, almost everything seemed normal, and there were no such issues to consider. However, some unexpected development in the foetus was suspicious of an early sign of alobar Holoprosencephaly. The condition was confirmed with the second sonographer based on the protocol guidelines.

References:

- Kim, A., Savary, C., Dubourg, C., Carré, W., Mouden, C., Hamdi-Roze, H., Guyodo, H., Douce, J.L. and Pasquier, L., 2019. Integrated clinical and omics approach to rare diseases: novel genes and oligogenic inheritance in holoprosencephaly. *Brain*, 142(1), pp.35-49.
- Monteagudo, A., 2020. Holoprosencephaly. *American Journal of Obstetrics & Gynecology*, 223(6), pp.B13-B16.
- Roessler, E., Hu, P. and Muenke, M., 2018, June. Holoprosencephaly in the genomics era. In *American Journal of Medical Genetics Part C: Seminars in Medical Genetics* (Vol. 178, No. 2, pp. 165-174).
- Dubourg, C., Kim, A., Watrin, E., de Tayrac, M., Odent, S., David, V. and Dupé, V., 2018, June. Recent advances in understanding inheritance of holoprosencephaly. In *American Journal of Medical Genetics Part C: Seminars in Medical Genetics* (Vol. 178, No. 2, pp. 258-269).
- Kruszka, P. and Muenke, M., 2018, June. Syndromes associated with holoprosencephaly. In *American Journal of Medical Genetics Part C: Seminars in Medical Genetics* (Vol. 178, No. 2, pp. 229-237).
- Ionescu, C., Calin, D., Navolan, D., Matei, A., Dimitriu, M., Herghelegiu, C., & Ples, L. (2018). Alobar holoprosencephaly associated with a rare chromosomal abnormality. *Medicine*, 97(29), e11521. doi: 10.1097/md.00000000000011521
- Medline Plus. (2022). Nonsyndromic holoprosencephaly: MedlinePlus Genetics. Retrieved 9 April 2022, from <https://medlineplus.gov/genetics/condition/nonsyndromic-holoprosencephaly/>

