

CASE REPORT

Fetal Ultrasonography of Holoprosencephaly

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ABSTRACT

Holoprosencephaly (HPE) occurs when the embryonic forebrain (prosencephalon) fails to divide into the cerebral hemispheres. Thus, resulting in a single-lobed brain structure with severe skull and facial defects. In most instances, the malformations are rather severe, and babies die before birth.

An HPE may be categorized into three variants (1) alobar, (2) semilobar, (3) lobar. Cyclopia a severe facial abnormality that is characterized by a single eye at the root of the nose with an associated supraocular proboscis (tubular nose) replacing the missing one.

Keywords: Cyclopia, Holoprosencephaly, Lobar, Proboscis, Single ventricle, Ultrasonography.

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INTRODUCTION

Holoprosencephaly is an uncommon condition which presents with severe abnormalities, and hence baby does not survive and usually dies before birth. Antenatal ultrasonography (USG) is the modality of choice.

CASE REPORT

In our case of a multigravida who presented for routine antenatal scanning revealed very classical findings of single ventricle, fused eyes, and proboscis (Figs 1 to 3). No sibling in the family has any similar abnormality.

Prenatal evaluation by means of three- (3D) and four-dimensional (4D) transabdominal USG was performed to identify alobar HPE (Fig. 4). Since no antenatal treatment could be instituted, after parental counseling it was decided to terminate the pregnancy.

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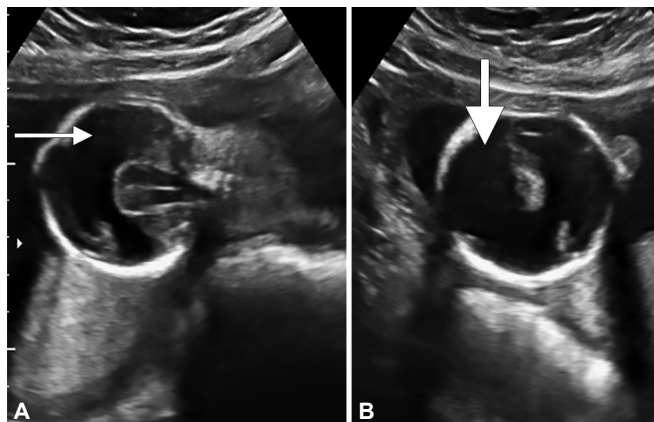
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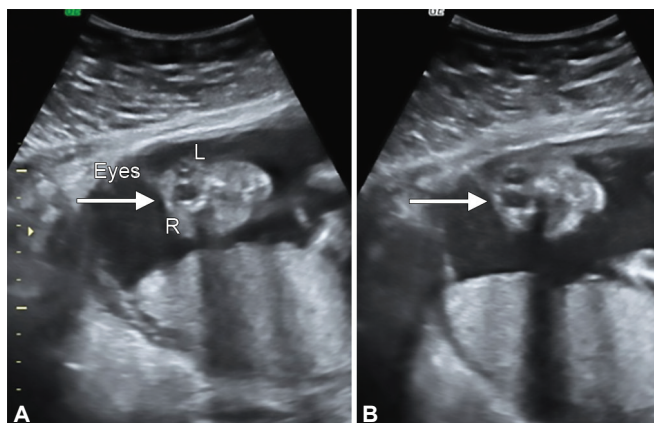
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DISCUSSION

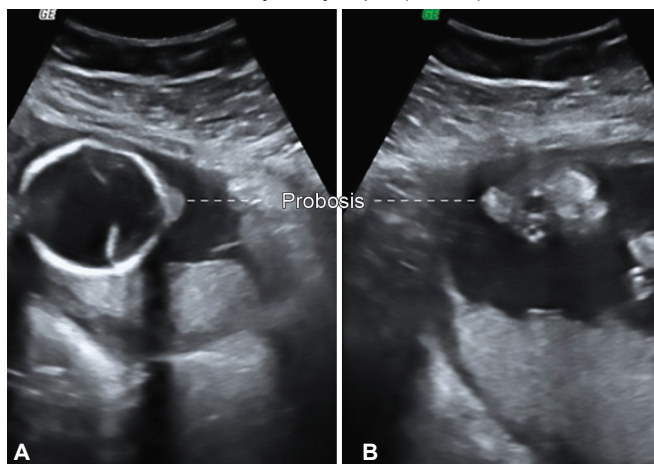
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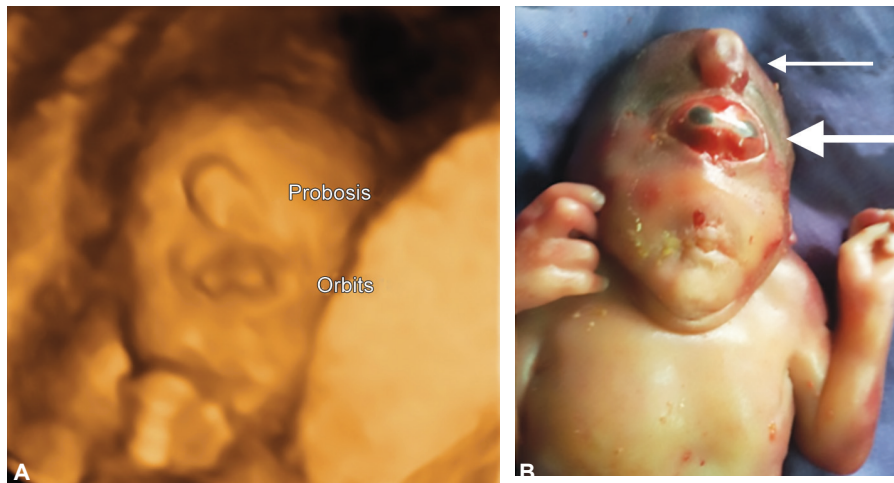
Figs 1A and B: Ultrasonography of fetus showing single ventricle: (A) Transverse (arrow) and (B) Longitudinal axis (arrow)



Figs 2A and B: Ultrasonography of fetus showing fused eyes "cyclops" (arrows)



Figs 3A and B: Ultrasonography of fetus showing absent nose and is seen in forehead as "proboscis" (arrows)



Figs 4A and B: (A) 3D Ultrasonography of fetus; (B) Aborted fetal face showing cyclops and proboscis (arrows)

nose with an associated supraocular proboscis (tubular nose) replacing the missing one. Alobar is often considered as the most serious form where the brain fails to cleave and is associated with facial anomaly and cyclopia. Semilobar is an intermediate form which is less severe where there is partial cleavage of the brain. In the lobar form, there is considerable evidence of a separate cerebral hemisphere noted and is the least severe form. In some instances of lobar holoprosencephaly, the patient's brain may be nearly normal.^{1,2}

Syntelencephaly which is sometimes also known as the middle interhemispheric variant results from the improper cleavage of posterior frontal and parietal lobes. If associated with cyclopia it is called cyclocephaly or synophthalmia and is considered a rare entity.^{2,3}

About 50% of the cases have a chromosomal abnormality most common being trisomy 13 (Patau) syndrome and trisomy 18 (Edwards) syndrome. There has been a recent increase in risk in infants born to diabetic mothers. Mutations in the gene encoding the sonic hedgehog protein, which is involved in the development of the central nervous system (CNS) has been attributed for. Some of the risk factors include toxoplasmosis, rubella cytomegalovirus, herpes (TORCH) infections, gestational diabetes, and history of previous miscarriages.¹⁻⁴

Some of the drugs deemed potentially unsafe during pregnancy, such as lithium, thiorazine, retinoic acid as well as anticonvulsants are said to have a contributory influence over the occurrence of HPE.^{5,6}

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