Quiz Section

Answer to Quiz No 3 (PMR Vol 1 No 03 September 2009)

1. What is the diagnosis?

Alobar Holoprosencephaly with proboscis (Ethmocephaly)

Holoprosencephaly is an error of ventral induction. It arises from disruption of the normal and patterning of the rostral neural tube during early embryogenesis. It is a spectrum of disorder resulting from absent or incomplete cleavage of the forebrain or prosencephalon, during early embryonic development in which a single embryological defect affects the growth of both brain and face.[1-5] Depending on the degree of cleavage of the prosencephalon, the condition is classified as alobar, semilobar or lobar.[1-5] If cleavage is absent, alobar type results with single ventricle, fusion of thalami and absent neurohypophysis and olfactory bulbs. The thin lining of ventricular cavity (thela choroidea) bulge out to occupy space between the calvarium and cerebral cortex, forming a CSF filled cyst called as dorsal sac. In semilobar type, partial cleavage occurs. The cerebral hemispheres are separated posteriorly and variable degrees of fusion of the thalami occurs, with associated absent olfactory bulb and corpus callosum. A single ventricle with rudimentary occipital horn is present. In lobar type, the hemispheres are separated anteriorly and posteriorly with certain degree of fusion of structures such as lateral ventricles and cingulate gyrus and absence of the cavum septum pellucidum. Incidence of these types varies from 1 in 1660 to 1 in 16000: while cyclopia or cebocephaly occurs in 1 in 4000 and 1 in 16000 births respectively.[1] Facial abnormalities always indicate abnormal brain, but may be absent even in severe cases, rarely.

All surviving individuals with more severe forms of holoprosencephaly have some developmental delay often persisting as mental retardation. Seizures, hypotonia or hypertonia, extrapyramidal features, swallowing difficulties, pituitary dysfunction and feeding difficulties are seen in the surviving individuals, Failure to thrive is very common[5]. In individuals with cyclopia or ethmocephaly, survival is rare beyond 1 week[5] Patients with lobar type, have normal life expectancy[1] If detected early, termination of pregnancy is advised especially in alobar and semilobar varieties. If detected later, conservative management and delivery is considered. Karyotyping should be done in all cases for the management of this and future pregnancies. Postnatal management involves appropriate medical specialities like perinatologist, clinical geneticist, endocrinologist, neurologist and craniofacial team for further diagnostic evaluation and therapy.[5]

References:

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