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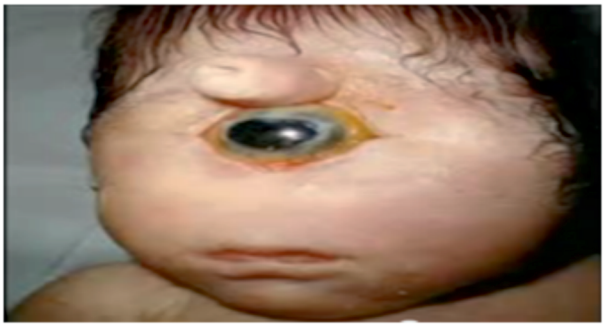
Holoprosencephaly/ Single Brain Ventricle

Mrs. Gauthami H M.Sc (N)
Asst Professor
Child Health Nursing Department
R V College of Nursing
Mob: 9035337411
Email ID: gauthamih.rvcn@rvei.edu.in

Abstract:

Holoprosencephaly (HPE) is a cephalic disorder condition in which the prosencephalon (the forebrain of the embryo) fails to develop into the two hemispheres. Normally, the forebrain is formed and the face begins to develop in the fifth and sixth weeks of human pregnancy.

The condition can be mild or severe. According to the National Institute of Neurological Disorders and Stroke (NINDS), in most cases of holoprosencephaly, the malformations are so severe that babies die before birth.



When theforebrain in the embryodoesn't divide to form bilateral cerebral hemispheres i.e.(the right and left halves of the brain), it causes defects in the development of thebrain structure, function & face .

In rare cases, babies are born with normal or near-normal brain development and facial anomalies that may affect the eyes, nose, and upper lip.

CAUSES:

The exact cause of Holoprosencephaly (HPE) is idiopathic (Not known).

Genetics:-

- The cause of cyclopia as a genetic malfunctioning during the process by which the embryonic brain is divided into two. Only the visual cortex take recognizable form, and at this point an individual with a single forebrain region will be likely to have a single large, eye (at such a time, individuals with separate cerebral hemispheres would form two eyes).
- Although many children with holopro-sencephaly have normal chromosomes, specific chromosomal abnormalities have been identified in some patients (trisomy of chromosome 13, also known as Patau syndrome).
- Sometimes HPE is inherited (autosomal dominant as well as autosomal or recessive inheritance).
- The features which are consistent usually with familial transmission of the disease (e.g., a single central maxillary incisor) should be assessed carefully in parents and family members.

Non-genetic factors:-

- Diabetes (Gestational),
- Transplacental Infections ("TORCH"),
- Bleeding in the first trimester

- History of any miscarriages.
- The disorder is found double the times in female kids.
- HPE and the use of various drugs are potentially unsafe for pregnant and lactating mothers as there is correlation between them.
- It includes birth control pills, insulin, lithium, aspirin, thorazine, retinoic acid and anticonvulsants.
- Alcohol consumption and HPE are also correlated alongwith nicotine, the toxins present in the cigarettes, when used during pregnancy.

CLASSIFICATION: -

There are four types of Holoprosencephaly.

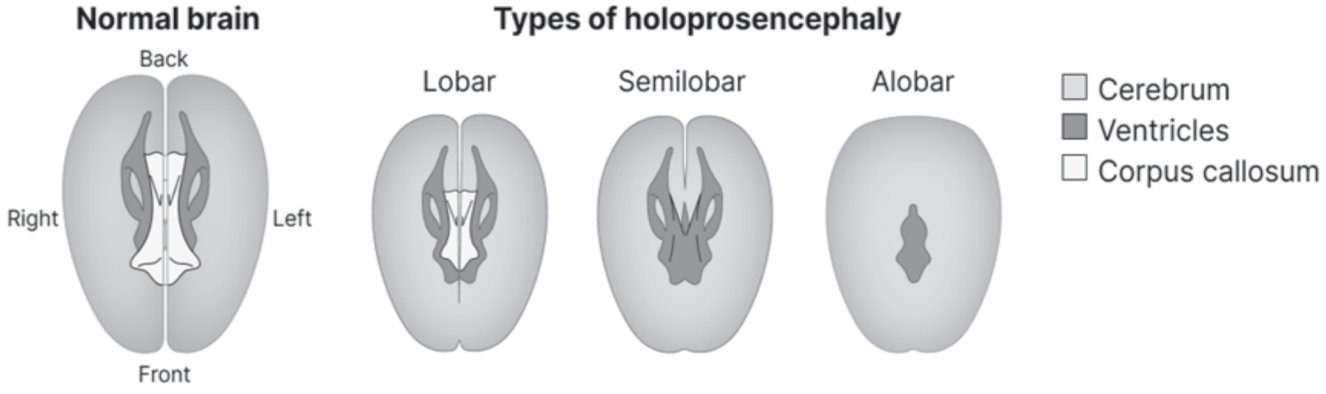
1. Alobar Holoprosencephaly
2. Semilobar Holoprosencephaly
3. Lobar Holoprosencephaly&
4. Middle Interhemispheric fusion variant (Syntelencephaly).

✓ Alobar Holoprosencephaly: It is the most severe form of holoprosencephaly where there isno separation of the cerebral hemispheres. It containssingle ventricle. The corpus callosum and interhemispheric fissures are absent and the thalami are usually unseparated.

✓ Semilobar Holoprosencephaly:The cerebral hemispheres separate only posteriorly.

✓ Lobar Holoprosencephaly: It is characterized by near complete separation of cerebral hemispheres except at frontal lobes.

✓ Middle Interhemispheric fusion variant (Syntelencephaly): It results from inseparableness of posterior frontal and parietal lobes.



SIGNS & SYMPTOMS:-

Clinical manifestations of HPE range from the mild (no facial/organ defects, anosmia, single central incisor) to moderate& moderate to severe (cyclopia) Holoprosencephaly are characterized by various craniofacial anamolies.

- The most severe craniofacial deformity is cyclopia with a single or partially divided eye in a single orbit with a proboscis above the eye and absent nose (arhinia).
- Ethmocephaly is characterized by hypotelorism with a proboscis between the eyes.
- Cebocephaly represents hypoteloric eyes with a single nostril nose

Other malformations include:

- Single central maxillary incisor,
- Cleft lip and palate (Midline or Bilateral),
- Flat nose,
- Absent nasal bridge,

- Microphthalmia,
- Absence of lateral philtral ridges,
- Absence of the superior lingual frenulum,
- Seizures,
- Infantile spasms,
- Patients often have feeding difficulties,
- Gastroesophageal reflux,
- Malnutrition,
- Pituitary dysfunction,
- Diabetes insipidus,
- Temperature dysregulation,
- Disturbed sleep-wake cycle,
- Recurrent respiratory tract infections,
- Chronic lung disease,
- Constipation
- Death is usually due to brainstem dysfunction or with infections and dehydration.

DIAGNOSIS: -

- ✓ A prenatal history should document possible environmental causes.
- ✓ A family history of miscarriages, terminations of pregnancy, neonatal, or infant deaths and craniofacial abnormalities is necessary.
- ✓ Prenatal ultrasound of the face and falx cerebri can be used to diagnose Alobar and Semilobar Holoprosencephaly as early as the first trimester.
- ✓ Fetal MRI provides more sensitive diagnosis for milder forms of holoprosencephaly during the third trimester.
- ✓ Genetic Testing
- ✓ EEG
- ✓ CT Scan
- ✓ MRI

MANAGEMENT: -

MEDICAL:-

- Anticonvulsants are used for treatment of seizures.
  Eg:Valproic acid, Phenytoin Carbamazepine, Lamotrigine, Zonisamide, Levetiracetam, Rufinamide, Ethotoin, Primidone, Meths uximide & Phenobarbital.
- Decarboxylase Inhibitors
  Eg: Levodopa/Carbidopa
- Anticholinergic Agents
  Eg: Benztropine, Trihexyphenidyl
- Skeletal Muscle Relaxants
  Eg: Baclofen
- Histamine H2 Antagonists
  Eg: Cimetidine, Ranitidine, Nizatidine, Famotidine
- Proton Pump Inhibitors
  Eg: Omeprazole, Lansoprazole, Pantoprazole
- Estrogens/Progestins
  Eg: Ethinyl estradiol and drospirenone Norethindrone acetate/ethinyl estradiol
- Spasticity, choreoathetosis, and dystonia may require symptomatic treatment.
- Hormone replacement therapy is used in cases of pituitary dysfunction.
- Promotility agents are used for Gastroesophageal reflux.

SURGICAL:-

- ✓ Ventriculoperitoneal shunt can be done if child has Hydrocephalus.
- ✓ For feeding difficulties, poor weight gain, and gastroesophageal refluxgastrostomy tube and Nissen fundoplication may be required

THERAPY OPTIONS FOR CHILDREN WITH HPE: -

It is a condition without a cure or standard course of treatment. Treatment for HPE is symptomatic and supportive. Some of the common therapy options for children with HPE are:

- ➢ ***Augmentative Communication (Alternative Communication (AAC) :-***

It is a method of communication used for

children and adults with severe speech and language disabilities

- ***Developmental Therapy: -***

It is aguided play for children where it creates a learning environment and activities for promoting skills in all areas of child's development. This therapy is child-directed. Therapists show parents the techniques used for behaviour management and modeling the context of everyday child experiences and environment.

- ***Hippotherapy:-***

It is a specialized form of physical therapy which uses a horse as a therapy tool to address movement disorders. This therapy is conducted by a licensed and specially trained health care provider, often a physical or occupational therapist or speech language pathologist. For the child with HPE and low muscle tone or poor motor control, the horse offers a movement experience that cannot be replicated in any clinic. The symmetric movements of the horse mimics human gait and provides neuromuscular information to the child's nervous system.

- ***Pediatric Feeding Therapy:-***

It is a combination of treatments which help the children who are having eating or drinking related difficulties. There are different types of feeding problems in children and the therapy is individualized for each child uniquely.

- ***Pediatric Occupational Therapy:-***

It focuses on child's sensory and motor skills, emotional development, self-care abilities and play skills to help improve successful functioning at home, school and community.

- ***Pediatric Physical Therapy:-***

It is provided by physical therapists who are licensed health care professionals with a master's or doctorate degree in physical therapy. A physical therapist assesses the areas concerned with joint motion, muscle control, strength, coordination, equilibrium, movement patterns, sensory issues, mobility, gait and equipment.

- ***Pediatric Speech and Language Therapy:-***

It provides assessment and treatment for a child with impaired communication skills including speech, language, play and social interaction development. The focus of treatment is on improving the oral-motor skills, developing the alternative ways for communication, improving language and cognitive skills.

- ***Vision Therapy:-***

It trains the entire visual system, which includes eyes, brain and body. The goal of it is to train the patient's brain to use the eyes for receiving the information effectively, comprehended quickly and react appropriately.

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