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# CHARGE SYNDROME

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**ABSTRACT**

CHARGE syndrome is a condition that is diagnosed based on clinical findings of coloboma, heart disease, choanal atresia, retarded growth or development, genital urinary anomalies and ear abnormalities or hearing loss. This syndrome is caused by genetic mutation of CHD7 gene[2]. This mutation disrupts the chromatin remodeling and gene expression which is responsible for sign and symptoms of CHARGE syndrome. The incidence of CHARGE syndrome is 1 in 8, 500 to 10,000 newborns[2]. An expert group classifies the major "4"C (Coloboma, choanal atresia, cranial nerve abnormality and Charge ear ) and minor criteria for diagnosis of Charge syndrome. Child's with all four or three major and three minor criteria most likely to have CHARGE syndrome. In coloboma it mainly affects retina and optic nerve and due to abnormality in cranial nerve child may have hearing loss and facial paralysis. Choanal atresia may be bilateral and unilateral or it may be bony or membranous choanal atresia. Congenital heart defect mostly tetralogy of fallot account 70-80% in child. Some children Develops behavioral and mental problems also. Children with charge syndrome require intensive medical and surgical interventions. They require continuous observation to assess the milestone. Child's with CHARGE syndrome require educational, occupational, speech therapy and behavioral therapies for to improve quality of life.

**Key words:** CHARGE, CHD7, Coloboma, Atresia , Syndrome.

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## INTRODUCTION

CHARGE syndrome is a rare autosomal dominant genetic disorder that arises during early fetal development and affects multiple organ systems. CHARGE syndrome is defined as a non-random association of anomalies (coloboma, heart defect, atresia choanae, retarded growth and development, genital hypoplasia, ear anomalies/deafness) [1]. The pattern of malformations varies among individuals with this disorder, and the multiple health problems can be life-threatening in infancy. Affected individuals usually have several major characteristics or a combination of major and minor characteristics [2]. In 1998, an expert group defined the major (the classical 4C's: Choanal atresia, Coloboma, Characteristic ears and Cranial nerve anomalies) and minor criteria of CHARGE syndrome. Individuals with all four major characteristics or three major and three minor characteristics are highly likely to have CHARGE syndrome.

Other name of this condition

- CHARGE association
- Hall-Hittner syndrome [2].

### Frequency

CHARGE syndrome occurs in approximately 1 in 8, 500 to 10,000 newborns [2].

### Causes

Typically caused by mutations in the chromodomain helicase DNA-binding protein-7 (*CHD7*) gene. This protein is found in many parts of the body before birth, including the eye, the inner ear, and the brain. In the brain, the CHD7 protein is active in several areas; including a bundle of nerve cells (neurons) called the olfactory bulb that is critical for the perception of odors. The CHD7 protein regulates the activity (expression) of several other genes through a process known as chromatin

remodeling. Most mutations in the *CHD7* gene lead to the production of an abnormal CHD7 protein that is broken down prematurely. Shortage of this protein is thought to disrupt chromatin remodeling and the regulation of gene expression. Changes in gene expression during embryonic development likely cause the signs and symptoms of CHARGE syndrome [2].

**SIGN AND SYMPTOMS**

CHARGE syndrome affects multiple organ systems, resulting in multiple problems apparent at birth. Other characteristics of CHARGE syndrome may not become apparent until later in life[4-5].

**Major symptoms (The 4 C's)**

COLOBOMA (an eye defect resulting in a keyhole shaped pupil and/or abnormalities in the retina or optic nerve). It is a cleft or failure to close of the eyeball during fetal development. This result in keyhole shaped pupil and abnormalities in retina, macula and optic nerve. Very small eyes (microphthalmia) or missing eyes (anophthalmia) can be severe forms of coloboma. Colobomas of the retina or optic nerve may result in significant vision loss, including blind spots, problems with depth perception or legal blindness. Colobomas occur most frequently in the retina and are present in at least 80-90% of patients with CHARGE syndrome.

**C: CRANIAL NERVE ABNORMALITIES**

✓ Abnormalities in Sensorineural (abnormalities in cranial nerve VIII), hypoplastic cochlea (81%) and absent of semicircular canal cause most common feature is hearing loss and difficulty in balance. The hearing loss can range from a mild hearing loss to profound deafness. CHARGE syndrome is associated with characteristic external ears that tend to protrude and lack lobes.

✓ Abnormality in cranial nerves IX and X cause most children have swallowing problems which include inability to coordinate suck and swallow, which results gagging and aspiration of food in lungs which leads pneumonia.

✓ Abnormalities in cranial nerve VII cause asymmetric facial palsy (paralysis of one side of face) resulting lack of facial expression.

✓ Abnormalities in cranial nerve I absent and reduce sense of smell that cause difficulty to learn eat normally.

**C:CHOANAL ATRESIA**

Choanae are the passages from the back of the nose to the throat which make possible to breathe through the nose. In 50% cases of CHARGE syndrome it is blocked (atresia) or narrowed (stenosis). It may be the unilateral and bilateral, which cause difficulty in breathe. If it is bilaterally present the newborn cannot breathe and there is a chance of respiratory failure, so immediate measure must be taken.

C: CHARGE ear

Most of the children with charge syndrome have unusual external ears. The "typical CHARGE ear" is short and wide with little or no earlobe. The helix (outer fold) may end abruptly in mid-ear. The center of the ear (concha) is often very triangular in shape. The ears are often floppy and may stick out due to weak cartilage. The two ears often look different from each other.

**Minor symptoms**

Symptoms which are less specific to CHARGE syndrome and not enough to be considered major i.e. heart defects, genital abnormalities, kidney abnormalities, cleft lip or palate, TE fistula or esophageal atresia, poor growth, hypotonia, typical CHARGE face, and typical CHARGE hand.

**Heart defects**

Approximate 75-80% children with CHARGE syndrome have congenital heart defect. The most common heart defect is tetralogy of fallot, VSD (Ventricular septal defect), AV canal defect and arotic arch anomalies. Severe heart defect are major cause of death in CHARGE syndrome.

**Genital abnormalities**

In boys with CHARGE syndrome have small penis, undescended testes (cryptorchidism) and abnormal urethral opening that is not at the end of penis (hypospadias). In girls the labia size is small.

**Kidney abnormalities**

Approximate in 40% of children with CHARGE syndrome have kidney abnormalities i.e. extra fluid in kidney (Hydronephrosis) , reflux of urine (backflow into kidney), small kidney, horseshoe kidney, multicystic dysplastic kidney.

**Cleft lip and/or cleft palate**

In children with CHARGE syndrome have a cleft lips and cleft palate; it is found approximate in 25% of children. The cleft lips may be one sided or two sided, may include the palate or not. Some children have cleft palate without cleft lips and some have sub mucous cleft palate (only mucus is cleft not involve the roof bone of mouth).

**Tracheoesophageal Fistula/Esophageal atresia**

About 15-20% of children with CHARGE syndrome have an abnormal opening between the trachea (air canal) and esophagus (food canal) and some child develop esophageal atresia (esophagus is not develop properly or may be constricted that why there is no connection between the stomach and esophagus).

**Hypotonia of the trunk**

Most of the children with CHARGE syndrome have lack of muscle tone of upper body especially the trunk that cause sloping shoulder. Due to this weakness delay in walking usually the child with CHARGE syndrome walks in 3to 4 year. This problem combined with balance or vision problem due to underdeveloped semicircular canal.

**Poor growth**

The birth weight of children normal but after some time birth weight decrease or children growth is affected all these due to the some nutritional problems or heart problem or other illness. A child's with CHARGE syndrome also shown growth hormone insufficiency.

**Typical CHARGE face**

The face of the child is very asymmetric. The typical child has a square face, with broad prominent forehead, arched eyebrows, large eyes, occasional droopy eyelids, a prominent nasal bridge with square root, small nostrils, prominent nasal columella, flat midface, small mouth, occasional small chin, which will improves with age.

**Typical CHARGE hand**

In typical CHARGE hand, there is a small thumb, broad palm with hockey sticks, palmer crease and short fingers.

**Other common feature**

Brain abnormalities (hydrocephalus, microcephaly), weak cry due to larngomalacia, some children have missing or extra nipples, wide neck, cervical vertebra abnormities , poor immune system. T- cell deficiency. Many children develop scoliosis due to weakness on upper part of body.

**Developmental feature**

Children with CHARGE syndrome shows delayed developmental especially due to sensory deficit (vision or hearing) and frequently illness or hospitalization. The child grows older shows the autistic behavior like hand waving or head banging and some child develop obsessive compulsive disorder also.

## DIAGNOSIS OF CHARGE SYNDROME

- All the suspected cases of CHARGE syndrome should have to complete physical examination to check the major and minor features of CHARGE syndrome. It includes heart (to find out any heart defect or abnormality), vision (to check the vision defect), hearing test (to check hearing loss) as well as intelligent and developmental test.
- Diagnosis made by the medical geneticist based on the presence of one major or several minor symptoms of CHARGE syndrome.
- Cranial CT scan ( to identify hypoplastic chochlea)
- MRI (brain, ear), Smell-Testing, kidney ultrasound, growth hormone stimulation test, immunological test and genetic testing [4, 5].

## TREATMENT

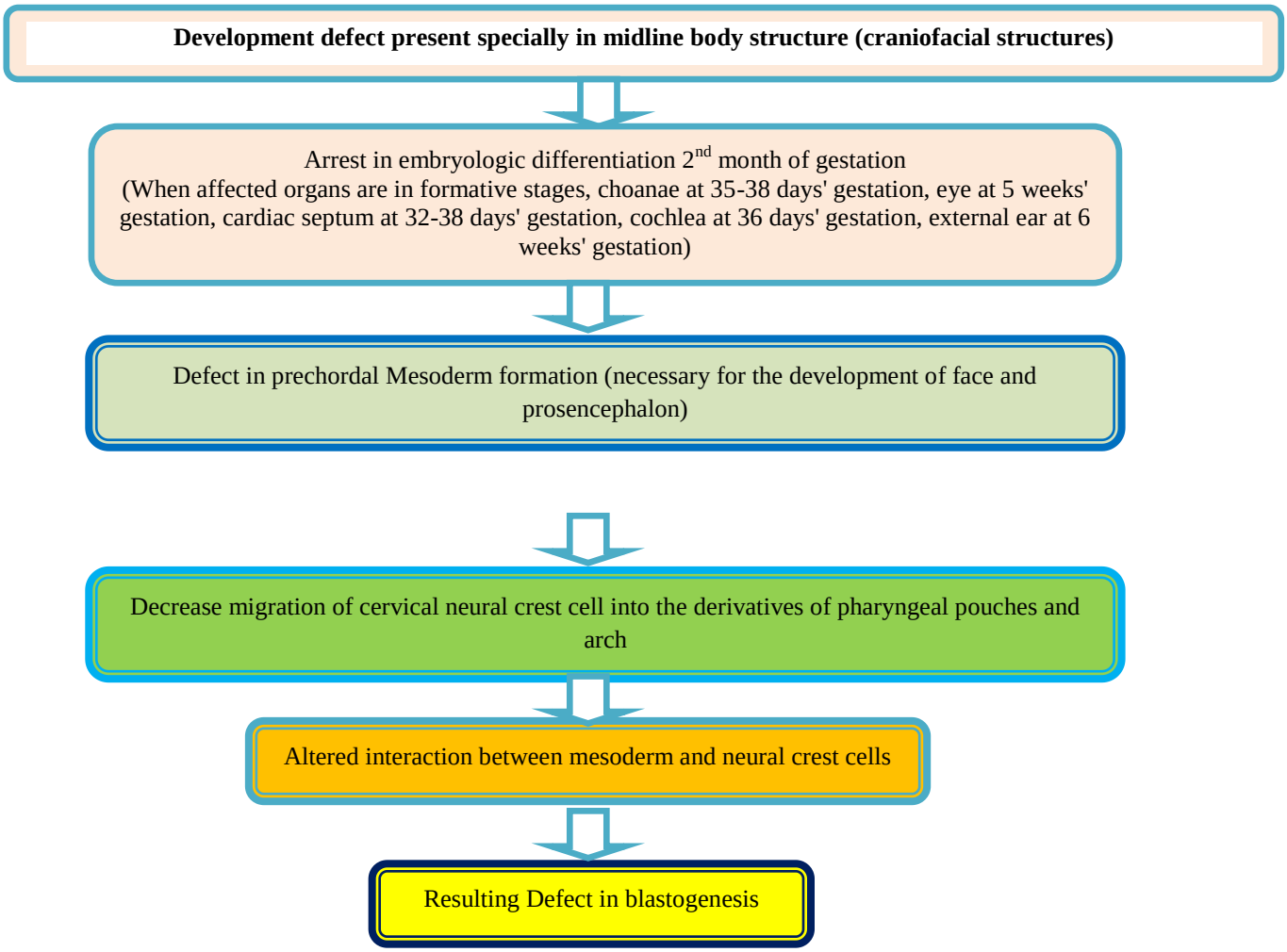
- Children with CHARGE syndrome require intensive medical as well as surgical management. The most common emergency in CHARGE syndrome baby is Cyanosis due to posterior and bilateral choanal atresia and congential heart defects. Thus primary focus of treatment is the airways stabilization and maintains circulation.
- Many congenital or structural abnormalities (heart defect, chonal atresia, cleft lip, cleft palate, esophageal fistula etc.) corrected surgically.
- Child with hearing loss receives cochlear implants to aid their sensorineural hearing loss.
- Problem with swallowing need to feed by the help of gastrostomy tube until they are not able to swallow safely.
- If child has Genital abnormality requires hormone therapy to achieve the puberty.
- Visual correction and language training start in early childhood.
- Genetic counseling, occupational therapy, speech therapy [5].

## NURSING RESPONSIBILITIES

The nursing care of a child with a CHARGE syndrome involves challenging task. Child with CHARGE syndrome present with several congenital disabilities and abnormalities. These children's require complete physical assessment of to find out the problem present in child and nursing assessment should be done on continuous basis for planning for nursing intervention. Nursing care of CHARGE syndrome is not specific, care is depending on the problem of the child. During neonatal period usually have breathing difficulty due congenital anomalies. The nurse has to maintain normal breathing pattern by maintaining position, administration of oxygen and suctioning if required. Assess the child for cyanosis and prevent cyanosis by administering oxygen to neonate if he has any cyanotic heart disease and choanal atresia.

Nurse has to monitor the vital signs and maintain IV chart and administer the medication as prescribed by physician and assist in diagnostic procedure. Feeding difficulty is one of the important nursing concerns; the nurse has to assess the nutritional status of the child and providing feeding via NG tube or Gastrostomy tube. Asses the milestone of the child and nurse has to help the parents to poster the normal growth and development of the child. Check the visual abnormalities and hearing loss. Main responsibility of nurse is to prevent injury and trauma due to these disabilities. Inform child and family members regarding child condition and obtain informed consent and assent from the child for required corrected surgery. Provide Preoperative care and postoperative care to the child who is undergoing any corrected surgery. Provide psychological and emotional support to family members to cope of with child condition and provide genetic counseling.

**Pathophysiology**




The complete function of *CHD7* during embryologic development remains unclear [3, 4].

## CONCLUSION

CHARGE syndrome is a genetic disorder, which is rarely seen in children. Mainly affect eyes, ear, respiratory tract, heart and child will have poor growth and developmental delay. The child with CHARGE syndrome requires multidisciplinary approach to treat and improve the quality of life.

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Nil

## CONFLICT OF INTEREST

No interest

## STATEMENT OF HUMAN AND ANIMAL RIGHTS

All procedures performed in human participants were in accordance with the ethical standards of the institutional research committee and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards. This article does not contain any studies with animals performed by any of the authors.

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