CHARGE SYNDROME

- A rare **pattern of malformations** affecting multiple organ systems
- Prevalence: **1 in 10,000 to 15,000** live births
- Typically **sporadic**; however, rarely, there may be an inherited genetic mutation or germline mosaicism

DIAGNOSIS

- Caused by a mutation in the ***CHD7* gene**, which plays an important role in embryonic development
- If clinical features of CHARGE, **genetic testing** is recommended to confirm the diagnosis

Coloboma

- Present in **70-90%**
- An eye abnormality, often a hole or gap in one of the structures of the eye, such as the iris, retina, or optic disc
- May lead to **vision impairment** or **blindness**, depending on the location and severity. Also, **at risk for glaucoma**.

Heart defects

- Present in **75-80%**
- Most common: atrial septal defects (**ASD**), ventricular septal defects (**VSD**), **tetralogy of Fallot, aortic arch anomalies**

Atresia choanae

- Present in **50-60%**
- **Obstruction of the nasal passages**, particularly the choanae (openings at the back of the nasal cavity)
- Can lead to **breathing difficulties** and **nasal congestion**

Restriction of growth and development

- Present in **70-80%**
- Both prenatal and postnatal **growth deficiency**
- **Developmental delays** can affect motor skills, speech, and cognitive abilities
- Early intervention and tailored educational support are crucial

Genital and urinary abnormalities

- Genital abnormalities may include underdeveloped or malformed genitalia; present in **60%**
- Urinary tract anomalies can vary and may include issues such as kidney abnormalities or structural problems in the urinary system; present in **40%**

Ear abnormalities and deafness

- Present in **> 90%**
- Malformations of the outer and middle ear – short and wide with little or no earlobe
- Hearing loss is common, and it can be conductive, sensorineural, or mixed

Some affected individuals may have other congenital malformations, including but not limited to, **microcephaly, cleft lip and palate, swallowing difficulties, cranial nerve abnormalities, tracheoesophageal fistula,** and **hypotonia.**

MANAGEMENT

Multidisciplinary healthcare team, including geneticists, pediatricians, surgeons, audiologists, SLP, OT/PT, and developmental specialists. Treatment emphasizes **developmental support** and **managing impairments**. Many of the structural abnormalities (choanal atresia, heart defects, cleft lip, TEF) can be surgically repaired.

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