

## Airway Clearance Indications: Chromosomal/Genetic Disorders

### CHARGE Syndrome

The term CHARGE is an acronym for an association of observed congenital (present at birth) defects that later came to be known as CHARGE syndrome. As understanding of the genetics of CHARGE syndrome has progressed, the hallmark clinical features of the disorder that resulted in the acronym are generally no longer relied upon to make the diagnosis, but the name remains.

CHARGE syndrome is a pleiotropic disorder. Pleiotropic disorders are disorders in which a single genetic mutation results in multiple, diverse defects. These disorders are complex and challenging, since associated defects may vary in expression and severity between individuals. Randomly occurring (not inherited) mutations in the CDH7 gene on chromosome 8 are thought to account for approximately 60-65% of all cases of CHARGE syndrome, although rare instances of inherited forms of the disorder have been reported. The incidence of CHARGE syndrome is approximately 1 in 8,500-10,000 births, with both genders and all races and ethnic groups equally represented. CHARGE syndrome is the leading cause of deaf-blindness at birth.

#### What Happens in CHARGE Syndrome?

The genetic mutation responsible for CHARGE syndrome results in numerous, diverse congenital defects. An observed pattern of common defects resulted in the acronym CHARGE, which stands for:

- **C** – Coloboma (hole or cleft) in the eye
- **H** – Heart defects
- **A** – Atresia of the choanae (blockage or narrowing of passage from the nose to the throat)
- **R** – Retardation of growth and/or development
- **G** – Genital and/or urinary defects
- **E** – Ear anomalies and/or deafness

It is important to note that not all patients with CHARGE syndrome will have all of the classic features and, in addition to the classic features, many individuals with CHARGE syndrome also exhibit other clinical or behavioral problems including immunodeficiency, hypotonia, cranial nerve abnormalities, gastroesophageal reflux, scoliosis, sinusitis and learning and communication disorders.

#### How Airway Clearance Therapy Can Help CHARGE Syndrome

Keeping the airways clear of excess secretions and thereby reduce the incidence of inflammation and/or infection and is crucial to maintaining respiratory health. Airway clearance therapy using High Frequency Chest Wall Oscillation (HFCWO) has been demonstrated by clinical study to promote excess mucus clearance and improve bronchial drainage. Shear forces are created by HFCWO treatment that mechanically releases adhered secretions from the walls of the



pulmonary tract. HFCWO has also been shown to reduce the viscosity of secretions which significantly improves mobilization of excess mucus. By replicating cough, HFCWO can effectively mobilize pulmonary secretions from smaller airways to larger airways where they can be coughed out, swallowed or suctioned.

**For More Information on CHARGE Syndrome:**

1. CHARGE Syndrome Foundation: <http://www.chargesyndrome.org/>
2. GeneReviews overview of CHARGE syndrome:  
<http://www.ncbi.nlm.nih.gov/bookshelf/br.fcgi?book=gene&part=charge>
3. CHARGE syndrome resource page from the National Organization for Rare Disorders (NORD):  
[http://www.rarediseases.org/search/rdbdetail\\_abstract.html?disname=CHARGE%20Syndrome](http://www.rarediseases.org/search/rdbdetail_abstract.html?disname=CHARGE%20Syndrome)
4. National Library of Medicine Genetics Home Reference page on CHARGE syndrome:  
<http://ghr.nlm.nih.gov/condition=chargesyndrome>