CHARGE Syndrome Fact Sheet

CHARGE syndrome (CS) refers to a specific set of birth defects, medical problems, and developmental issues. The most distinctive birth defects are coloboma, choanal atresia and characteristic ears (external ears and small/absent semicircular canals).

- **Diagnosis** should be made by a Medical Geneticist. Diagnosis is based on key features, ideally with DNA testing for *CHD7* mutations. Key features:
  - Coloboma
  - Cranial nerve abnormalities
  - Choanal atresia
  - Heart defects
  - Characteristic external ears
  - Esophageal defects
  - Small/absent semicircular canals
  - Genitourinary abnormalities
  - *CHD7* gene mutations

- **Incidence**: One in every 8-10,000 births. Every person with CS has a unique set of features. There is wide variation in physical features and cognitive ability.

- **Cause**: Mutations in the *CHD7* gene on chromosome 8 are found in 80-90% of cases. There is no relationship to sex, race, nationality, religion or socio-economic status or prenatal exposures.

- **Recurrence**: It does not usually run in families. Recurrence risk to unaffected parents is 1-2%. If a parent has CS, risk to a baby is 50/50.

- **Sensory deficits**: Most individuals with CS have difficulty with hearing, vision and balance. This results in delayed motor development and communication. The educational term for combined vision and hearing deficits is “deafblind.”

- **Cognitive ability & testing**: Many have decreased cognitive abilities, but 30-50% have normal intelligence. Intelligence of children with CS is often underestimated due to the effects of combined hearing, vision and balance issues. Testing, therapies and educational intervention MUST take into account hearing, vision and balance status.

- **Lifespan**: There is an increased mortality, especially in the first two years. Although individuals with CS remain medically fragile, lifespan can be normal.

- **Outcome**: Individuals with CS need medical care appropriate to their particular features. In addition, early intervention and appropriate and challenging educational and vocational programs specific to their sensory needs are imperative. Although there are many problems, children with CS can survive and become healthy, happy citizens.