

## Examples of Rare Disorders with Congenital Anomalies

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### Description

Congenital anomalies, commonly known as birth defects are structural or functional anomalies that occur during intrauterine life. These anomalies can be identified prenatally, at birth, or sometimes may only become apparent later in life. While some congenital anomalies are common and well-studied, others are rare, often associated with genetic disorders that affect only a small fraction of the population. These rare congenital anomalies pose unique challenges for diagnosis, management, and research due to their low prevalence and the complexity of their manifestations. Congenital anomalies can be classified into two main categories: Structural anomalies and functional/ developmental anomalies. Structural anomalies involve problems with the formation of body parts, such as heart defects, cleft lip and palate or spina bifida. Functional anomalies involve problems with how a body part or system works, which can include metabolic disorders, sensory impairments and intellectual disabilities. Rare disorders, also known as orphan diseases are defined as conditions that affect a small percentage of the population. In the United States, a disease is considered rare if it affects fewer than 200,000 people. Despite their rarity, there are thousands of rare disorders, many of which have genetic origins and are associated with a wide range of congenital anomalies.

### Examples

**Rett syndrome:** Rett syndrome is a rare genetic neurological disorder that predominantly affects females. It is caused by mutations in the *MECP2* gene located on the X chromosome. The disorder typically manifests after an initial period of normal development, followed by a regression in motor and cognitive abilities. Congenital anomalies associated with Rett syndrome include microcephaly (small head size), scoliosis and growth failure. Affected individuals often exhibit stereotypic hand movements and severe intellectual disability.

**Ehlers-Danlos syndrome:** Ehlers-Danlos syndrome is a group of connective tissue disorders characterized by hypermobility of joints, skin hyperextensibility and tissue fragility. EDS is caused by mutations in various genes responsible for collagen production and structure. Congenital anomalies in EDS can include heart valve defects, vascular abnormalities and gastrointestinal issues. The severity of symptoms and anomalies varies widely among affected individuals.

**Williams syndrome:** Williams syndrome is a rare genetic disorder caused by the deletion of genetic material on chromosome 7. It is characterized by distinctive facial features, cardiovascular abnormalities, and developmental delays. Common congenital anomalies in Williams syndrome include supravalvular aortic stenosis (narrowing of the large blood vessel carrying blood from the heart), kidney anomalies and gastrointestinal issues. Individuals with Williams syndrome often have a unique cognitive profile with strengths in verbal abilities and sociability.

### Diagnosis and management

The diagnosis and management of congenital anomalies in rare disorders present several challenges:

**Limited awareness and knowledge:** Due to their rarity, many healthcare providers may have limited experience and knowledge about these disorders. This can lead to delays in diagnosis and appropriate management. Increasing awareness and education among healthcare professionals is essential for improving outcomes.

**Genetic testing and counseling:** Genetic testing plays a vital role in diagnosing rare disorders and understanding their underlying causes. However, access to genetic testing and counseling can be limited, particularly in resource-poor settings. Ensuring equitable access to these services is essential for accurate diagnosis and family planning.

**Multidisciplinary care:** The complexity of congenital anomalies in rare disorders often requires a multidisciplinary approach involving various specialists, including geneticists, cardiologists, neurologists, orthopedic surgeons and developmental pediatricians. Coordinating care among these specialists can be challenging but is essential for comprehensive management.

**Treatment development:** Research on rare disorders is often limited by funding constraints and the small number of affected individuals. Despite these challenges, advances in genomics and personalized medicine for understanding and treating rare disorders. Collaborative efforts among researchers, healthcare providers and patient advocacy groups are essential for advancing knowledge and developing effective treatments.