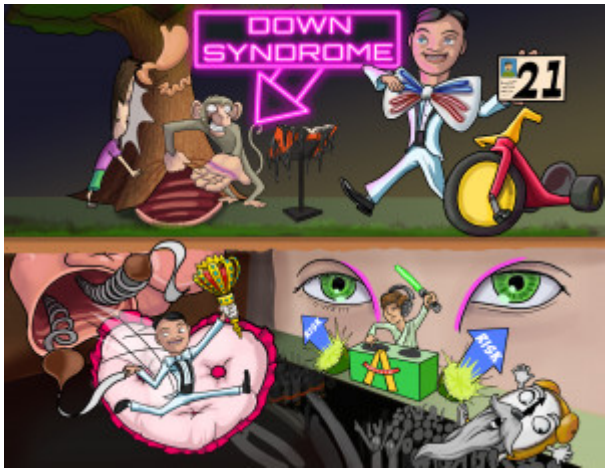
Down Syndrome

Down syndrome is caused by the presence of all, or part of a third copy of chromosome 21, secondary to meiotic nondisjunction (95%), Robertsonian translocation (4%), or Down mosaicism (1%). Down syndrome is the most common chromosomal disorder, and the most common genetic cause of mental retardation. It is characterized by a variety of dysmorphic features, congenital malformations, and predisposition to other medical conditions. Characteristic dysmorphic features include flat facies, prominent epicanthal folds, simian crease, and gap between first and second toes. Congenital malformations include endocardial cushion defects, with the most common being a septum primum type atrial septal defect and duodenal atresia. Patients with Down syndrome also have a greater predisposition for Alzheimer's dementia and acute lymphoblastic leukemia.



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Pathophysiology

Trisomy 21
Tricycle (21) ID
The presence of three copies of chromosome 21 due to meiotic nondisjunction, Robertsonian translocation, or Down mosaicism.

Meiotic Nondisjunction
Extra Chromosome Bow-tie
The failure of chromosome pairs to separate properly during meiosis 1 and 2.

Signs and Symptoms

Intellectual Disability (Mental Retardation)
Tar Covered Book
Significantly impaired cognitive functioning and deficits in two or more adaptive behaviors.

Single palmar crease
Simian with Simian Crease
A single line that runs across the palm of the hand.

Flat Facies
Flatten Face
Patients with Down syndrome have characteristically flat facies consisting of a low nasal bridge and a small nose.

Duodenal Atresia
Duodenal Atresia-tree
Duodenal atresia is a birth defect of the digestive tract where the duodenum is not patent. This occurs more frequently in infants with Down syndrome. Duodenal atresia is commonly diagnosed with a “double bubble” sign on imaging.

Hirschsprung's Disease
Hershey-spring
This disease is characterized by the loss of Auerbach's plexus in the gut, causing failure of a segment of the colon to relax. Hirschsprung's disease is more common in individuals with Down syndrome.

Septum Primum Type ASD
ASD Scepter
While the majority of atrial septal defects are septum secundum type, Down syndrome is most commonly associated with a septum primum type ASD.

Endocardial Cushion Defects

Heart Cushion

A range of congenital cardiac defects that are characterized by involvement of the atrial septum, the ventricular septum, and one or both of the AV valves.

Prominent Epicanthal Folds

Epicanthal Folds

A prominent skin fold of the upper eyelid, covering the inner corner medial canthus, is common in Down syndrome.

Increased Risk of Acute Lymphoblastic Leukemia

Up-arrow Risk of Acute-angle Lime-blasting Leukemia-Luke

Children with Down syndrome have a markedly increased risk for acute lymphoblastic leukemia.

Alzheimer's Disease

Old-timer

People with Down syndrome begin to have symptoms of Alzheimer's disease in their late 40s or early 50s. Although the exact cause is unknown, the early onset of Alzheimer's may be linked to the extra copy of chromosome 21, because it can lead to increased production of beta-amyloid in the brain.