

	<b>Down Syndrome</b>	<b>Cerebral Palsy</b>
<b>Description</b>	A chromosomal abnormality in chromosome number 21 which results in characteristic facial features, mental retardation, and other physiological manifestations.	A group of neuromuscular disorders, often resulting from brain damage suffered during development and early growth, resulting in movement, sensory, and neurologic impairments.
<b>Etiology</b>	<p>Chromosomal subtypes:</p> <ul style="list-style-type: none"> <li>• Trisomy 21: approximately 95% have an extra 21 chromosome.</li> <li>• Translocation: approximately 3-4% have a portion of the extra 21 chromosome break off during cell division and attach itself to either chromosome 14, 21, 22.</li> <li>• Mosaicism: approximately 1% have a failure of separation of the 21<sup>st</sup> chromosome after fertilization resulting in some cells having 46 chromosomes and others having 47 chromosomes.</li> </ul>	<ul style="list-style-type: none"> <li>• Preconception <ul style="list-style-type: none"> <li>○ Parents' biological age</li> <li>○ Environmental toxins</li> <li>○ Malnutrition for parents</li> <li>○ Radiation damage</li> </ul> </li> <li>• Prenatal <ul style="list-style-type: none"> <li>○ Endocrine Dysfunction</li> <li>○ Malnutrition</li> <li>○ Toxins: alcohol, smoking</li> <li>○ Infection</li> <li>○ Placental pathology</li> <li>○ Prematurity</li> <li>○ Rh incompatibility</li> <li>○ Hypoxia</li> <li>○ Cerebral hemorrhage</li> </ul> </li> <li>• Natal <ul style="list-style-type: none"> <li>○ Mechanical respiratory obstruction</li> <li>○ Maternal anoxia/hypoxia</li> <li>○ Breech delivery</li> <li>○ Prolapse of cord</li> <li>○ Cerebral hemorrhage</li> <li>○ Prematurity</li> </ul> </li> <li>• Postnatal <ul style="list-style-type: none"> <li>○ Cerebral hemorrhage</li> <li>○ Encephalitis</li> <li>○ Meningitis</li> <li>○ Anoxia/hypoxia</li> </ul> </li> </ul>
<b>Incidence</b>	About 1 in every 700 live births	1.5-4 individuals for every 1000 live births
<b>Signs/ Symptoms</b>	<ul style="list-style-type: none"> <li>• Craniofacial <ul style="list-style-type: none"> <li>○ Microcephaly</li> <li>○ Round head with flattened back of skull</li> <li>○ Flat facial profile</li> <li>○ Upward slanting of eyes/Epicanthic fold (inner corner eyes are rounded)</li> <li>○ Micrognathia</li> <li>○ Narrow palate</li> <li>○ Delayed eruption of teeth with missing teeth</li> <li>○ Areas of missing hair</li> <li>○ Ear canals are narrow</li> </ul> </li> <li>• Cardiac <ul style="list-style-type: none"> <li>○ Atrioventricular canal defects</li> <li>○ Septal defects</li> <li>○ Mitral valve prolapse</li> </ul> </li> <li>• Gastrointestinal <ul style="list-style-type: none"> <li>○ Esophageal and duodenal atresia</li> <li>○ Annular pancreas</li> <li>○ Complications related to vomiting, constipation, failure to thrive, feeding difficulties, and intolerance</li> </ul> </li> <li>• Musculoskeletal <ul style="list-style-type: none"> <li>○ Hypotonus</li> <li>○ Joint laxity/hyperflexibility of joints</li> <li>○ Atlanto-axial instability</li> <li>○ Hip dysplasia</li> <li>○ Instability of the knee cap</li> <li>○ Flat feet</li> <li>○ Scoliosis</li> </ul> </li> <li>• Cognitive <ul style="list-style-type: none"> <li>○ Mental retardation</li> <li>○ Increased incidence of Alzheimer's disease</li> </ul> </li> <li>• Respiratory <ul style="list-style-type: none"> <li>○ Respiratory tract infection</li> <li>○ Acute and chronic airway obstruction</li> <li>○ Sleep apnea</li> </ul> </li> <li>• Other <ul style="list-style-type: none"> <li>○ Delayed growth</li> <li>○ Vision loss/Hearing loss</li> <li>○ Middle ear infections</li> <li>○ Hypothyroidism</li> <li>○ Obesity</li> <li>○ Seizure disorders</li> </ul> </li> </ul>	<ul style="list-style-type: none"> <li>• Symptoms are related to areas within the brain that are effected.</li> <li>• Common symptoms include: <ul style="list-style-type: none"> <li>○ Spasticity</li> <li>○ Paralysis</li> <li>○ Seizures</li> <li>○ Muscle spasms</li> <li>○ Contractures</li> <li>○ Irregular breathing <ul style="list-style-type: none"> <li>▪ Excessive rate of breathing</li> <li>▪ Decrease in vital capacity</li> </ul> </li> <li>○ Delayed development of motor skills</li> <li>○ Mental retardation</li> <li>○ Dysarthria</li> <li>○ Apraxia</li> </ul> </li> <li>○ Dysphonia</li> <li>○ Visual abnormalities</li> <li>○ Hearing abnormalities</li> <li>○ Perseveration</li> <li>○ Reduced mobility</li> <li>○ Malnutrition</li> <li>○ Gastrointestinal manifestations <ul style="list-style-type: none"> <li>▪ Impaired visceral motility</li> <li>▪ Gastroesophageal reflux disease (GERD)</li> <li>▪ Abdominal pain</li> <li>▪ Chronic constipation</li> </ul> </li> </ul>

<p><b>Swallowing Manifestations</b></p>	<ul style="list-style-type: none"> <li>• Anterior tongue rest posture with anterior open bite</li> <li>• Hypotonia of muscles of mastication, oral preparation, oral transfer</li> <li>• Tongue hypotonia</li> <li>• Hypotonic soft palate</li> <li>• Difficulty with initiation of suck</li> <li>• Weak lip seal</li> <li>• Poor bolus control and transit</li> <li>• Tongue thrusting</li> <li>• Decreased effectiveness of chewing</li> <li>• Difficulty coordinating suck/swallow/breathe</li> <li>• Drooling</li> <li>• Increased incidence of respiratory infections</li> </ul>	<ul style="list-style-type: none"> <li>• Increased anxiety during feeding for both caregiver and the individual with cerebral palsy (Parrot, et al.; 1992)</li> <li>• Anterior tongue rest with anterior open bite</li> <li>• Altered occlusion</li> <li>• Food refusal</li> <li>• Tongue thrust</li> <li>• Tongue retraction</li> <li>• Lip retraction</li> <li>• Tactile hyper- or hypo- sensitivity</li> <li>• Oral aversion</li> <li>• Hyper- or hypo- tonicity of oral/pharyngeal/laryngeal musculature</li> <li>• Primitive reflexes <ul style="list-style-type: none"> <li>○ Rooting</li> <li>○ Bite</li> <li>○ Gag <ul style="list-style-type: none"> <li>▪ Hypoactive</li> <li>▪ Hyperactive</li> </ul> </li> <li>○ ATNR</li> </ul> </li> <li>• Pathologic reflexes and motor patterns <ul style="list-style-type: none"> <li>○ Bite</li> <li>○ Use of tongue to assist with forming seal around edge of cup for sucking</li> </ul> </li> <li>• Anterior loss of food from oral cavity</li> <li>• Impaired initiation of suck</li> <li>• Impaired oral bolus formation/control/transit</li> <li>• Impaired suck/swallow/breathe synchrony</li> <li>• Decreased pharyngeal peristalsis</li> <li>• Airway compromise <ul style="list-style-type: none"> <li>○ Decreased efficiency and timeliness of laryngeal movement</li> <li>○ Hypertonicity supra- and infra- hyoid muscles</li> <li>○ Coughing</li> <li>○ Choking</li> <li>○ Throat clearing</li> <li>○ Aspiration/penetration</li> </ul> </li> <li>• Impaired postural control <ul style="list-style-type: none"> <li>○ Impaired head neck control</li> <li>○ Impaired jaw stability</li> <li>○ Impaired sitting balance</li> <li>○ Use of postural changes to assist with swallowing</li> </ul> </li> <li>• Drooling</li> <li>• Fatiguing during feeding/decreased endurance</li> <li>• Respiratory distress during oral feeding <ul style="list-style-type: none"> <li>○ Hypoxia during swallow</li> <li>○ Upper respiratory infections (URI)/pneumonia</li> <li>○ Increased respiratory load</li> <li>○ Impaired esophageal motility</li> </ul> </li> </ul>
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