

	Rett Syndrome	Spina Bifida
Description	An inherited syndrome, primarily affecting females, characterized by characteristic mental and physical degeneration after a period of normal development.	Congenital disorder where vertebrae and spinal column do not close before birth.
Etiology	Genetic basis	<ul style="list-style-type: none"> • Hereditary linked • Folic acid deficiency • Virus
Incidence	1 in 9000 in girls under age of 12; 1 in 30,000 in general population	Between 1 and 10 infants per 1000 births
Signs/ Symptoms	<ul style="list-style-type: none"> • During first approximately 5 months the individual is developmentally on track. • Slowing of head growth • 30% reduction in brain size • Loss of previously acquired hand skills • Stereotypic hand movements <ul style="list-style-type: none"> ○ Wringing ○ Squeezing ○ Waving ○ Hand mouthing/thumb sucking • Growth retardation • Athetosis • Rigidity/spasticity • Mental retardation • Seizures • Dystonia • Bradykinesia • Reduction in eye contact • Delays in gross motor development • Constipation • Breathing abnormalities <ul style="list-style-type: none"> ○ Breath holding ○ Apnea ○ Hyperventilation ○ Aerophagia: swallowing of air ○ Arrhythmic breathing pattern 	<ul style="list-style-type: none"> • Alterations in sleep patterns • Gait abnormalities • Apraxia • Scoliosis • Impaired communication skills • Gastroesophageal reflux disease (GERD) • Vertebrae do not completely form • Cerebral palsy • Partial or complete paralysis • Loss of bowel and bladder control • Mental retardation • Hip dislocation
Swallowing Manifestations	<ul style="list-style-type: none"> • Teeth grinding • Anterior tongue rest posture with anterior open bite • Alterations in temporomandibular joint musculature • Altered occlusion • Lingual retraction • Tongue thrusting • Impaired suck • Impaired chewing • Impaired suck/swallow/breathe synchrony <ul style="list-style-type: none"> ○ Uncontrolled spillage of bolus from oral cavity into pharyngeal cavity ○ Pooling in valleculae and pyriform sinuses ○ Breath holding during swallow ○ Initiation of swallowing on in-breath 	<ul style="list-style-type: none"> • Impaired bolus formation/control/transit • Increased hypertonicity of infra- and supra-hyoid musculature • Impaired airway protection <ul style="list-style-type: none"> ○ Delayed pharyngeal swallow ○ Aspiration/penetration • Primitive reflex patterns <ul style="list-style-type: none"> ○ Rooting ○ Bite
		<ul style="list-style-type: none"> • Decreased pharyngeal sensation and/or motility • Decreased esophageal sensation and/or motility • Achalasia • Dyspraxia • Impaired gastrointestinal motility