List of Rare Diseases and Chronic Illnesses:

Achondroplasia: is a common cause of dwarfism. It is a genetic disorder. People with achondroplasia have short stature, with an average adult height of 52 inches for males and 48 inches for females. The disorder causes an abnormality of cartilage formation. The prevalence is approximately 1 in 25,000.

Batten's Disease: is a rare, fatal neurodegenerative disorder that begins in childhood. Early symptoms of the disorder usually appear around ages 2–10, with gradual onset of vision problems, or seizures. Early signs may be subtle personality and behavior changes, slow learning or regression, repetitive speech or echolalia, clumsiness, or stumbling. There may be slowing head growth in the infantile form, poor circulation in lower extremities (legs and feet), decreased body fat and muscle mass, curvature of the spine, hyperventilation and/or breath-holding spells, teeth grinding, and constipation. Over time, affected children suffer mental impairment, worsening seizures, and progressive loss of sight, speech and motor skills. Eventually, children with Batten disease become blind, bedridden, and demented, and die. Batten Disease is a terminal disease; life expectancy varies depending on the type or variation.

Cerebral Palsy: is a general term for a group of permanent, non-progressive movement disorders that cause physical disability, mainly in the areas of body movement. There may also be problems with sensation, depth perception, and communication ability. Difficulty with cognition and epilepsy are found in about one-third of cases. Cerebral palsy is caused by damage to the motor control centers of the developing brain and can occur during pregnancy, during childbirth, or after birth up to about age three.

Ciliary Dyskinesia: also known as **immotile ciliary syndrome**, is a rare genetic disorder that causes a defect in the action of the cilia lining the respiratory tract (lower and upper, sinuses, Eustachian tube, and middle ear) and fallopian tube, and also of the flagella of sperm in males. The main consequence of impaired ciliary function is reduced or absent mucus clearance from the lungs, and susceptibility to chronic recurrent respiratory infections, including sinusitis, bronchitis, pneumonia, and otitis media.

Congenital Heart Defects: or **congenital heart anomaly** is a defect in the structure of the heart and great vessels that is present at birth. Many types of heart defects exist, most of which either obstruct blood flow in the heart or vessels near it, or cause blood to flow through the heart in an abnormal pattern. Other defects, such as long QT syndrome, affect the heart's rhythm. Heart defects are among the most common birth defects and are the leading cause of birth defect-related deaths. Approximately 9 people in 1000 are born with a congenital heart defect. Many defects do not need treatment, but some complex congenital heart defects require medication or surgery.

Cystic Fibrosis: is a genetic disorder that affects most critically the lungs, and also the pancreas, liver, and intestine. It is characterized by thick, viscous secretions. Difficulty breathing is the most serious symptom and results from frequent lung infections that are treated with antibiotics and other medications. Other symptoms—including sinus infections, poor growth, and infertility—affect other parts of the body. The main signs and symptoms of cystic fibrosis are salty tasting skin, poor growth and poor weight gain despite normal food intake, accumulation of thick, sticky mucus, frequent chest infections, and coughing or shortness of breath.

Hunter Syndrome: is a serious genetic disorder that primarily affects males. It interferes with the body's ability to break down and recycle specific mucopolysaccharides (sugars). The symptoms of Hunter syndrome are generally not apparent at birth, but usually start to become noticeable after the first year of life. Often, the first symptoms of Hunter syndrome may include abdominal hernias, ear infections, runny noses, and colds. Since these symptoms are quite common among all infants, they are not likely to lead a doctor to make a diagnosis of Hunter syndrome right away. Physical appearances of many children with Hunter syndrome include a distinctive coarseness in their facial features, including a prominent forehead, a nose with a flattened bridge, and an enlarged tongue. For this reason, unrelated children with Hunter syndrome often look alike. They may also have a large head as well as an enlarged abdomen. Many continue to have frequent infections of the ears and respiratory tract.

Hypoplastic Left Heart Syndrome: is a complex and rare heart defect present at birth (congenital). In hypoplastic left heart syndrome, the left side of the heart is critically underdeveloped. If a baby is born with hypoplastic left heart syndrome, the left side of the heart can't effectively pump blood to the body, so the right side of the heart must pump blood both to the lungs and to the rest of the body. Medication to prevent closure of the connection (ductus arteriosus) between the right and left sides, followed by either surgery or a heart transplant, is necessary to treat hypoplastic left heart syndrome.

Idiopathic Infantile Pyoderma Gangrenosum: is a condition that causes tissue to become necrotic, causing deep ulcers that usually occur on the legs. When they occur, they can lead to chronic wounds. Ulcers usually initially look like small bug bites or papules, and they progress to larger ulcers. Though the wounds rarely lead to death, they can cause pain and scarring. The little girl who has this is 1 of 17 children in the entire world with this condition.

Maple Syrup Disease – also called branched-chain ketoaciduria is a genetic disorder where the body is unable to process certain protein building blocks (amino acids). The name comes from the sweet odor of affected infants' urine, and the smell can also be in the ear wax. Infants with this disease seem healthy at birth but if left untreated suffer severe brain damage and eventually die. From early infancy, symptoms of the condition include poor feeding, vomiting, dehydration, lethargy, hypotonia, seizures, hypoglycemia, ketoacidosis, pancreatitis, coma and neurological decline. Maple syrup urine disease affects approximately 1 out of 180,000 infants, and occurs most commonly in the Amish and Mennonite population.

Metatropic Dysplasia – This is a genetic mutation characterized by dwarfism, enlarged joints, problems with vision and hearing. The word "metatropic" is derived from the Greek word "metatropos" meaning "changing form". Clinically, this is one that progresses over time. Affected individuals develop curvature of the spine and degenerative arthritis. Because it is so uncommon, the exact incidence is not known.

Mitochondrial Disease – This is a group of diseases caused my dysfunctional mitochondria (a subunit within the cell) that generates energy. They are most commonly caused by mutations in the DNA. There are many specific types of mitochondrial diseases. The symptoms are usually poor growth, loss of muscle coordination and weakness, visual and hearing problems, learning disabilities, heart, liver, kidney, gastrointestinal, and respiratory, as well as other neurological problems. About 1 in 4,000 children in the United States will develop mitochondrial disease by the age of 10 years. Up to 4,000 children per year in the US are born with a type of mitochondrial disease. Because mitochondrial disorders contain many variations and subsets, some particular mitochondrial disorders are very rare.

Osteogenesis Imperfecta – This is known as the "brittle bone disease", which is a congenital bone disorder characterized by brittle bones that are prone to fracture. These patients have a defect in the connective tissue, and unable to make collagen. Patients also have curvature of the spine, loose joints, discoloration of the sclera (whites of the eyes) and sometimes hearing loss. Severe cases also have respiratory issues. Diagnosis of OI is based on the clinical features and may be confirmed by collagen or DNA testing. There is no cure for OI. Treatment is aimed at increasing overall bone strength to prevent fracture and maintain mobility. The incidence of OI is estimated to be 1 per 20,000 live births.

Primordial dwarfism - is a form of dwarfism that results in a smaller body size in all stages of life beginning from before birth. More specifically, primordial dwarfism is a diagnostic category including specific types of profoundly proportionate dwarfism, in which individuals are extremely small for their age, even as a fetus. Most individuals with primordial dwarfism are not diagnosed until they are about 3 years of age. It is rare for individuals affected by primordial dwarfism to live past the age of 30.

Short gut syndrome - Also known as short bowel syndrome is a malabsorption disorder caused by the surgical removal of the small intestine, or rarely due to the complete dysfunction of a large segment of bowel. Most cases are acquired, although some children are born with a congenital short bowel. Symptoms can include abdominal pain, diarrhea, fluid depletion, weight loss, easy bruising and fatigue. Patients are unable to absorb vitamins and minerals.

Spinal Muscular Atrophy – SMA is a disease caused by a genetic defect in the survival motor neuron 1gene. SMN1 is apparently selectively necessary for survival of motor neurons, as diminished abundance of the protein results in death of neuronal cells in the anterior horn of the spinal cord and subsequent system-wide muscle wasting (atrophy). SMA manifests in various degrees of severity, which all have in common general muscle wasting and mobility impairment. Other body systems may be affected as well, particularly in early-onset forms. SMA is the most common genetic cause of infant death.

Wolf-Hirschhorn - This is a genetic condition caused by partial deletion of the short arm chromosome 4. Wolf-Hirschhorn syndrome is a condition that affects many parts of the body. The major features of this disorder include a characteristic facial appearance, delayed growth and development, intellectual disability, and seizures. Antibody deficiencies are also common. Severity of symptoms and expressed phenotype differ based on the amount of genetic material deleted. The prevalence is estimated to be 1 in 50,000 births. However, this may be an underestimate because it is likely that some affected individuals are never diagnosed. For unknown reasons, Wolf-Hirschhorn syndrome occurs in about twice as many females as males.