



List of 10 rare diseases affecting children

In support of Rare Disease Day on 28 February, what follows is a list of only 10 of the thousands of rare diseases that affect children. Children diagnosed with any of these 10 diseases would benefit from a palliative care approach.

1. **Batten Disease** affects boys and girls. Symptoms of Batten disease usually start between the ages of 5 and 10 years and include loss of vision or seizures. Over time there is a loss of muscle control and some wasting of brain tissue. Progressive sight loss and dementia occur. There is no treatment available to cure or slow the progression of Batten disease and it is always fatal, with death usually in the late teens or early twenties. [Learn more](#)
2. **Duchenne muscular dystrophy (DMD)** affects the use of voluntary muscles in the body and is inherited, primarily affecting boys of all ethnic backgrounds. Normal development occurs initially but between the ages of 2 and 6 the affected child may have difficulty walking, running or climbing and struggle to lift their head due to a weak neck. Eventually the heart and breathing muscles are affected which leads to difficulty breathing, fatigue and heart problems due to an enlarged heart. Even with the best medical treatment young men with DMD seldom live beyond their early thirties. [Learn more](#)
3. **CANDLE Syndrome** (Chronic Atypical Neutrophilic Dermatitis with Lipodystrophy and Elevated Temperature Syndrome) is a very rare auto inflammatory disease. It is an inherited, genetic condition. Patients have recurring fevers, beginning in infancy, which happen almost daily. They also present with delayed development, skin rashes and unique facial features such as thicker lips, swollen eyelids. Children develop swelling around the eye sockets, clubbing of fingers and toes and gradual enlargement of the liver. There is not effective therapeutic treatment for CANDLE syndrome and life expectancy is compromised with death often resulting from organ inflammation. Quality of life is also severely affected. [Learn more](#)

4. **Childhood Interstitial Lung Disease or chILD**, is a broad term for a group of rare lung diseases that can affect babies, children and teens. The disease harms the lungs by damaging the tissues that surround the alveoli and bronchial tubes and sometimes the air sacs and airways. Lung function is decreased, blood oxygen levels reduced and the breathing process is disturbed. The disease has only been researched in the last decade and it is not known how many children have each type of chILD. Severity differs according to the type of the disease but can lead to early death. There is no cure. [Learn more](#)
5. **Ehlers-Danlos syndromes** are a group of genetic disorders which share common features including easy bruising, joint hypermobility, skin that stretches easily and weakness of tissues. Symptoms vary in severity according to the form of the disorder and treatment according to the particular manifestations present in the patient. Symptoms may also affect the autonomic nervous system used for breathing and urination. [Learn more](#)
6. **Ellis Van Creveld syndrome** is an inherited disorder due to an error on Chromosome 4 and is usually diagnosed at birth. Symptoms include short stature, short forearms and legs, extra fingers and toes, narrow chest with short ribs and malformed pelvis. 50 – 60% have a heart defect. Respiratory infections are common and about half those born with this syndrome die in early infancy. [Learn more](#)
7. **Gaucher disease** (Types 1, 2 and 3) is an inherited storage disorder where fatty substances build up to toxic levels in the spleen, liver, lungs, bone marrow and sometimes in the brain. It is genetically inherited and affects both boys and girls. Symptoms of Gaucher Type 2 begin in infancy, usually by 3 months and these children seldom live past 3 years of age. [Learn more](#)
8. **Krabbe Disease** has 4 subtypes, each beginning at different ages. Type 1 is the most common and begins between 3 – 6 months. It affects the nerve cells and causes nerve cell damage, leading to loss of use of muscles, increasing muscle tone, arching of the back and damage to vision and hearing. There is no cure or way to stop the disease once it is in full swing and babies with the Type 1 infantile form usually die by 13 months. [Learn more](#)
9. **Neuroblastoma** is a rare and aggressive childhood cancer of unknown cause. It usually affects children under the age of five, and can occur before a child is born, often spreading to other parts of the body before any symptoms become apparent. Long-term survival for children with advanced disease older than 18 months of age is poor and most of the survivors have long-term effects from the treatment. [Learn more](#)
10. **Pompe disease** is caused by a deficiency or lack of an enzyme, leading to the build-up of glycogen and has an infantile and late onset form. The former usually appears in the first few months of life where babies have trouble holding up their heads. The heart muscles become diseased and the heart becomes enlarged and weak. Babies with the infantile form usually die before their first birthday due to heart failure and respiratory weakness. [Learn more](#)

Learn more at www.rarediseaseday.org