Recently, a question was sent to me asking about certain types of muscular dystrophy and what the symptoms might be. The most prevalent form of MD is Duchenne’s, and it affects hundreds of thousands of young children and adults.

Duchenne muscular dystrophy (DMD) is a genetic disorder characterized by progressive muscle degeneration and weakness. It is one of nine types of muscular dystrophy. DMD is caused by an absence of dystrophin, a protein that helps keep muscle cells intact. Symptom onset is in early childhood, usually between ages 3 and 5. The disease primarily affects boys, but in rare cases, it can affect girls.

Muscle weakness can begin as early as age 3, first affecting the muscles of the hips, pelvic area, thighs and shoulders, and later the skeletal (voluntary) muscles in the arms, legs and trunk. The calves often are enlarged. By the early teens, the heart and respiratory muscles also are affected.

Becker muscular dystrophy (BMD) is a milder version of DMD. Its onset is usually in the teens or early adulthood, and the course is slower and less predictable than that of DMD.

Duchenne muscular dystrophy was first described by the French neurologist Guillaume Benjamin Amand Duchenne in the 1860s, but until the 1980s, little was known about the cause of any kind of muscular dystrophy. In 1986, MDA-supported researchers identified a particular gene on the X chromosome that, when flawed (mutated), leads to DMD. In 1987, the protein associated with this gene was identified and named dystrophin. Lack of the dystrophin protein in muscle cells causes them to be fragile and easily damaged.

DMD has an X-linked recessive inheritance pattern and is passed on by the mother, who is referred to as a carrier. DMD carriers are females who have a normal dystrophin gene on one X chromosome and an abnormal dystrophin gene on the other X chromosome. Most carriers of DMD do not themselves have signs and symptoms of the disease, but a minority do. Symptoms can range from mild skeletal muscle weakness or cardiac involvement to severe weakness or cardiac effects and can begin in childhood or adulthood.

Until relatively recently, boys with DMD usually did not survive much beyond their teen years. Thanks to advances in cardiac and respiratory care, life expectancy is increasing and many young adults with DMD attend college, have careers, get married and have children. Survival into the early 30s is becoming more common, and there are cases of men living into their 40s and 50s.

Children with Duchenne muscular dystrophy (DMD) often are late walkers.

In toddlers, parents may notice enlarged calf muscles. This enlargement is known as pseudohypertrophy, or “false enlargement,” because the muscle tissue is abnormal and may contain scar tissue.

A preschooler with DMD may seem clumsy and fall often. Parents also may note that children have trouble climbing stairs, getting up from the floor or running.

By school age, children may walk on their toes or the balls of their feet with a slightly waddling gait, and fall frequently. To try to keep their balance, they may stick out their bellies and pull back their shoulders. Children also have difficulty raising their arms.

Many children with DMD begin using a wheelchair sometime between ages 7 and 12. Transition to a wheelchair usually is a gradual process; at first, the chair may be required only to conserve the child’s energy when covering long distances. (Children often experience renewed independence once they fully transition to a power wheelchair.)

In the teen years, activities involving the arms, legs or trunk may require assistance or mechanical support.

The Muscular Dystrophy Association provides help and hope to the families affected by MD by providing counseling, support and all the services they need when faced with this diagnosis.