Dear Dad,

I'm so sorry to hear that Mr. and Mrs. Friedson's son, Brian, has been diagnosed with Duchenne Muscular Dystrophy. Muscular Dystrophy is an inherited disease that causes a person's muscle fibers to be highly susceptible to damage, and over time, these muscles become weaker. Duchenne Muscular Dystrophy, in particular, is the most severe of the nine types of muscular dystrophies, and it affects voluntary muscles (muscles used to purposefully move body parts), the heart, and breathing muscles.

As is the case with most DMD sufferers, Brian is male and was diagnosed early in his life (most are diagnosed between the ages of 2 and 6). He exhibited general weakness in his hips, pelvis, thighs, and shoulders, and his calves were enlarged. He fell frequently and had difficulty getting up, running, and jumping. Mr. And Mrs. Friedson said that he had a waddling-like gait and wondered if he suffered from mild mental retardation. He was officially diagnosed with DMD following genetic testing and a muscle biopsy (where some of his muscle cells were removed and observed with a microscope). Prior to that, the doctors may have developed a suspicion of muscular dystrophy using blood tests, ultrasound, and electromyography.

The majority of Duchenne Muscular Dystrophy patients are male because DMD is a recessive trait carried on the X chromosome. Chromosomes are the small units of genetic material that carry traits, and half of a person's chromosomes come from each of their parents. Thus, each person has two copies of genetic material for each trait. Recessive traits, like DMD, are only observed if both copies of the gene carry the recessive trait. However, the genes that determine gender, the X and Y-chromosomes, carry different genes. Males have one X and one Y-chromosome, but females have two X-chromosomes. Thus, if a male gets the recessive trait on the X-chromosome from his mother, he will express it. Females, on the other hand, will only express it if both copies of the trait (one X-chromosome from her mother and one X-chromosome from her father) are recessive.

Duchenne Muscular Dystrophy is caused by the absence of functional dystrophin, which is the result of this recessive trait. Dystrophin is a protein (a functional molecule in the body) that is needed by muscle cells for structural support. More specifically, dystrophin helps to anchor the internal cellular cytoskeleton (which is like a skeleton for the cell) to the surface membrane of the cell. Without dystrophin, the cell membrane, which is usually only permeable to water and specific other substances, allows other things to pass through as well. When this foreign matter enters the cell, the cell increases in internal pressure and explodes, which destroys the muscle cell-by-cell, essentially piece-by-piece.

DMD will probably kill Brian in his late teens or early 20s from problems with his heart or lungs. Although there is no cure, current medications (including prednisone) and therapies can slow the progression of the disease. In addition, current research on mice is promising and may lead to the development of more effective therapies in the future.

Best wishes,
David

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