

10 Facts about Duchenne (doo-shen) Muscular Dystrophy:

- 1.** Duchenne is classed as a rare disease, affecting 1 in 3,600 males. It affects 300,000 worldwide, with 54 boys born with Duchenne worldwide every single day.
- 2.** Duchenne is a disorder of the Dystrophin gene, the largest protein-coding gene in the entire human genome. There are 79 exons on the Dystrophin gene, just one mutation in any one of these exons is all it takes to cause Duchenne.
- 3.** Those with Duchenne are unable to produce Dystrophin, a protein essential for the stability and repair of muscle cells. Without it, every muscle in the body will eventually waste away.
- 4.** Those with Duchenne usually appear healthy at birth, but often struggle to meet developmental milestones. They struggle to run and climb stairs like healthy children and fall often. The typical age of diagnosis is around 4-5 years old.
- 5.** Walking ability is typically lost between 8-12, with a strong decrease in arm function generally occurring between 10-16. Those with Duchenne are not expected to reach their 30th birthday, in fact complications can mean some don't even reach their 20th.
- 6.** Unlike most genetic diseases, Duchenne occurs across ALL races and nationalities.
- 7.** Not all cases of Duchenne are passed on by carrier females. At least a third of cases are caused by a new genetic mutation with no previous family history. Quite simply, it could happen to any family.
- 8.** Females can have Duchenne too. Carriers can be manifesting, meaning they display some symptoms of Duchenne, varying from muscle pain and fatigue to Duchenne of the same severity as seen in males. The odds of a female having Duchenne of the same severity as boys is 1 in 50 million. Extremely rare, but it does happen.
- 9.** Those with Duchenne often have unusually large calves, this is known as pseudo-hypertrophy. It's caused by dying muscle cells being replaced by fat and scar tissue.
- 10.** There is currently NO effective treatment for Duchenne.

www.parentprojectmd.org www.cureduchenne.org