

**PATIENTS  
ARE  
WAITING...**

*Messages from Duchenne Muscular Dystrophy Families to the FDA*

The FDA's job is to make important and often challenging decisions that determine access to potential treatments. Many of these decisions rely on qualitative assessments that are based on quantitative data, balancing patient need and opportunity, speed and certainty, and benefits and risks. In some cases it may be simple for FDA experts to imagine what is in the best interest of the end user.

But in cases of rare, progressive diseases that are 100% fatal, it is a serious challenge for those who haven't "lived it" to anticipate the best interests of patients and families. Here, the Duchenne and Becker muscular dystrophy community provides some answers that are as impassioned and important as the community itself.

A program of:

**Parent Project  
Muscular Dystrophy**

LEADING THE FIGHT TO END DUCHENNE



# "PATIENTS ARE WAITING..."

## Messages from Duchenne Muscular Dystrophy Families to the FDA

To collect meaningful information from a broad group of people and caregivers managing Duchenne muscular dystrophy (Duchenne) and to advance the development of new treatments, Parent Project Muscular Dystrophy (PPMD) launched the **Benefit/Risk in Duchenne Therapies** program in late 2012. The goal is to inform the FDA and other government agencies, as well as biopharmaceutical companies about the priorities and risk tolerance of the Duchenne community.

Duchenne families often feel as if the FDA is an untouchable and unreachable group of professionals tasked with making critical decisions on potential drugs. In order to bridge that gap, PPMD's Benefit/Risk Program includes:

1. A rigorous first-of-its kind benefit risk study, that produced data on treatment preferences and risk tolerance using stated-preferences methods, and
2. "Share Your Story," an open-ended survey that allowed parents and patients to speak frankly to the FDA.

### Objectives and Methods

The program objective is to share stories from the community with the FDA and other stakeholders to help them better understand the perspectives of Duchenne families. Using an online, open-ended survey implemented on the Parent Project Muscular Dystrophy website, we asked families, "If you had a chance to talk to the FDA, what would you want them to know?" No further prompts were given to participants. The responses were publically available, so participants could read previous entries before posting their own comments.

We developed a codebook of recurring themes and coded each story individually. We then identified major and supporting themes from the entire set of stories and chose representative quotations. Readers should keep in mind that, based on the open-ended nature of the question posed, one might not expect extremely high concordance among stories that were spontaneously generated (without prompts).

Data is vitally important to inform the FDA's benefit-risk assessments. But nothing can replace the impact of sharing family stories of their experiences of living with Duchenne muscular dystrophy.

### Results

163 people participated in Share Your Story—a mix of individuals with Duchenne, parents of individuals with Duchenne, brothers and sisters, uncles and aunts, and grandparents. Quotations and stories are included from families who consented to their information being shared with the FDA and others.

### Major Themes

Participants had several key messages that they listed as the **most important** things they wanted to communicate to FDA leadership.

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#### **BURDEN OF DUCHENNE**

Reported by >90% of participants

The most common theme reflected on the **burden of managing Duchenne**, on patients, caregivers, the family, and the broader community. Specific burdens include **progressive loss of function and ability to participate** for the patients; **anticipating further disease progression and a short lifespan; impact on caregivers, siblings, and other relatives**; and



**need for treatments, services, and supports** that are unavailable. Representations of the burden of Duchenne can be found in the full stories provided at the end of this document.

## 2 URGENT NEED

Reported by 78% of participants

The large majority of participants described an **urgent need** based on **time running out** for us/our children. Under that theme, many participants expressed a **compelling, immediate need for access to clinical trials and new treatment options**.

*"All of us as parents know what the outcome is if we don't try something. I beg you to fast track drugs and treatments through the Duchenne pipeline. Time is something our boys do not have."*

*"Because DMD shortens the life span of those afflicted, I would encourage the FDA to consider being more aggressive with the approval of certain drugs where the benefits*

*include quality of life and life span. Some families welcome the risks that may come with treatments, feeling that doing something is better than just waiting for their child to deteriorate and die."*

*"We hear each tick of the clock very loudly in our heads, worried that science will take too long to develop a treatment that will slow/eliminate the progression. Our bigger worry is that science will develop it and the FDA will take too long to approve it."*

## 3 INCREASED FLEXIBILITY

Reported by 62% of participants

To meet the urgent need for new treatment options, more than half of the participants urged the FDA, sponsors, and other stakeholders to come together and **facilitate a faster, more flexible drug development and approval process**. This major theme includes requests to **harmonize efforts between U.S. and other regulatory bodies** to allow

access to all people with Duchenne, regardless of where they live.

*"Unfortunately, we don't see the FDA as an organization that protects us from harmful products/treatments, but we see them as an obstacle to progress. An obstacle to treatment being developed in time for my son, so that he does not become a statistic of lives lost to DMD."*

*"Simply put, we don't have years to wait. While I understand the need for FDA approval, and the concerns over possible risks and side effects - we aren't overly concerned with what these drugs could potentially do to our boys in the future. Without the medications, these boys have no future."*

#### **4 EXPANDED ACCESS**

Reported by 48% of participants

About half of the participants requested **expanded access to experimental and off-label drugs** for any Duchenne muscular dystrophy patient who chooses to use the drug. This major theme included a desire for sponsors and regulators to be **more inclusive of a broader range of patients in clinical trials**.

*"One aspect of this process I find most challenging is the limitation and regular exclusion of non ambulant individuals. My sons were willing participants and may have been ideal candidates for other studies, yet they were ineligible because they were unable to complete a six minute timed walking test used as a primary outcome measure. This narrow thinking is unacceptable as it excludes a significant number of patients and barring any progress in therapy, all ambulant patients will lose the ability to walk."*

#### **5 RISK TOLERANCE**

Reported by 28% of participants

Many participants communicated that **some risk is tolerable and individuals should have a choice**. These participants were willing to accept risk for

access to drugs, even experimental or off-label drugs. Parents expressed that they should be in control of decisions made for their children. Several expressed frustration that decisions regarding acceptable levels of risk are in the hands of FDA scientists who do not understand their lived experience.

*"When it comes to terminal illnesses [the FDA's] job should be to make sure a product is safe and that the risks and benefits presented by the producer are accurate. Our job should be to determine, given all that information, whether to give it to our children. It is an intensely personal decision that involves the parents and the child with Duchenne."*

*"He is dying anyways, what other choice do I have as a parent then let him take a drug that hasn't been through the rig-a-roll of getting years of research? We as DMD parents have no options. It's die or die trying..."*

#### **6 HOPE FOR A BETTER FUTURE**

Reported by 24% of participants

From a positive perspective, about a quarter of participants expressed that they have **hope for a better future** because of current and future clinical trials.

*"But there is now hope for our boys with Duchenne, hope for a brighter future. Research is starting to show results, more drugs are being developed to treat Duchenne. With more potential drugs there is more hope that my son and hundreds of other young men like him, will live full and rewarding lives standing on their own two feet."*

# STORIES

Out of the hundreds of stories, we chose several that seemed to reflect the most common sentiments.



**MILES**, 6 years old

## Left Waiting

Like many parents have, I'll start by giving my son's age. Miles is 6 years old and in 1st grade. The significance of age is obvious to all of us. It is a critical marker of where he is in a journey with a tragic arc, where every day, month and year counts. Miles is a happy kid. At 6, he is able to walk and play, participate in most activities at school and at home, dress himself, travel, ride the bus and swim in the ocean. That's not to say he doesn't have challenges - he has a cognitive delay, has never been able to run or jump, and is now old enough to start to feel the difference between his abilities and those of other kids. Still, life is sweet, for now. I believe that is partly because as parents we have striven to convey a sense of optimism and excitement in life. As every child should, he is growing up with expectation of a bright future. I wish we could freeze time and stay in our current reality forever.

That brings me to what I dread. Miles doesn't yet know that in many ways, life is (may be?) better now than it will ever be for him again. I dread the day that brutal reality sinks in to his consciousness-just the recognition of inevitable, brutal, crushing decline and loss of abilities feels unbearable. I would do almost anything to prevent or postpone that moment of terrible recognition. But, I know that without help, it's coming-maybe in a year or two. Each day that passes will see him lose a little more of his capability, mobility, stamina. My heart goes out to all the parents who have sat with their child through that horrible moment or series of small realizations that amount to the same. I know that wouldn't be the end - life and hope and joy would still go on. And, I know this is an old story that keeps repeating itself in family after family. Now we can break that cycle. Thanks to the incredible breakthroughs in medical research, we can change the story for many of our boys! There is hope for a much brighter future... miraculously there but maddeningly just out of our reach. Miles is incredibly lucky to have an exon deletion that makes him a candidate for 2 potential competing drugs that are in trial. He was in one of the potential drugs early trials on a low dose arm. Everyone, not just his parents but researchers, family and friends, saw him get stronger with the drug. For the first time in his life, he was running and (almost) jumping. We believe it also affected his learning - while we can't be 100% sure of the cause, during the trial his language skills made a big jump. It was a wonderful time of hope and progress. It's been over 6 months since the trial ended. He hasn't jumped or run since, and seems more tired than he did then. We understand the questions about efficacy of this potential drug but we sincerely believe that this happened. Miles would also be a candidate for the other potential drug. We are ready to go wherever and whenever we need to get it for him. We would drop our jobs, sell our house, move to another country. Because we know what will happen if he doesn't get the medication, we know



**CHRISTOPHER**, 12 years old

it's terrible, we know every passing day is a closed door. And we're just like all the parents with kids in this position. Every day that we delay costs our sons. Sure, my son can hang on for a year, he'll probably still be walking in 2 or 3. Longer? Well, we understand what abilities he will lose - it's the natural progression of DMD. FDA, I AM BEGGING YOU TO MAKE THE TREATMENTS AVAILABLE AS QUICKLY AND AS WIDELY AS POSSIBLE! CHANGE THE PROTOCOLS. BE FLEXIBLE. MAKE THIS HAPPEN. We can't wait. I'd like to hear from the FDA what parents and advocacy groups can do to provide the cover and support they need to alter their standard approach. Thank you, Elliott Barnett

### **Race Against Time**

My name is Mindy Cameron and my son Christopher was diagnosed with Duchenne in 2003 when he was two years old. He does not have one of the more common gene mutations and his disease is

progressing very typically, according to what we know from Duchenne natural history. I find that we often hear of the boys who defy the natural history of Duchenne. We marvel at the young men who still walk at age 17 and beyond, we talk of 30 somethings who hold down jobs and even have families, we get so excited about boys who are able to drive or go to college. But those boys are not typical. The reality is that most boys continue to lose ambulation around age 12 and die in their early 20s. My son will be 13 in July 2014 and he lost what little ambulation he had been able to hang on to about a month ago when he fell and broke his leg. It has been a devastating transition for our family, even though we are trying to hide the pain and focus on all of the new freedoms that he has in his power wheelchair. In truth, I hate the power wheelchair and everything that it represents. I know that we are now in the second of what is described as the three phases of Duchenne. I know that permanent seating brings on a host of health





**ELIJAH**, 9 years old

complications, like contractures and scoliosis and weight gain and increased heart rate and decreased pulmonary function. He is 12 years old. He should be running and growing and instead he is changing seating positions and using the standing function on his power chair. He is lifted onto the toilet and unable to get into his best friend's house because they have steps. He is sitting on the railroad tracks and the train is coming full speed ahead. I am one of those parents who have tried anything and everything to keep a step ahead of Duchenne and now I feel like I have lost a 10-year fight. The expensive supplements and medications, the physical therapy sessions for which insurance won't pay, and the night time routines that I subject my son to have made no difference in the relentless pace of his condition. His growth is stunted and his behavior has been affected by the toxic steroids that I've given him since he was 3-years-old and he struggles socially because of it. I feel like a failure as a parent for putting him through it all for what feels like nothing. The only interventions that we have for Duchenne are nowhere near good enough. We need to develop and approve treatments in the absolute fastest way possible. The FDA needs to use every means available to get help to these boys. The Duchenne community has been very assertive in expressing its willingness to take risks in light of the absolute horrific nature of this disease with no good treatment options. We are currently waiting for an FDA decision about the first exon skipping therapies for some Duchenne boys. These first drugs may not be perfect and our means to demonstrate their effectiveness may be incomplete, but these first drugs give every indication of effectiveness and

they should be given the green light while we work on improvements. You cannot get back what this disease takes from boys every day. The loss of a step in a timed test indicates something much much more than loss of function. It is death itself and it is coming for these boys at an insidious pace. I beg the FDA to intervene and act quickly for these children. We pay close attention to what is given priority review and accelerated approval and breakthrough status. We are at a loss as to why our boys do not seem to be a priority, when we have some incredibly promising technologies that could transform this disease. Maybe it is too late for my Christopher, but I still hold onto hope that it is not.

### **Willing to Accept Risk, Hope for Exon Skipping**

My son, Elijah, is 9 years old. His favorite thing to do is help me in the kitchen. He particularly enjoys baking. Other things he likes to do include dancing, drawing, and crafts. When he was four years old, he was diagnosed with DMD. As the diagnosis was not new to me; I was devastated. I knew that it was a slow death sentence. I have a family history and watched my brother, David, slowly lose muscle strength. This began with the legs, then progressed to all the other muscles in the body, his body deteriorated before my very eyes, and then finally his heart muscle failed at the age of 18. I know that without a viable therapy, I will slowly lose my son to this devastating diagnosis. As the disease progresses, he will no longer be able to complete basic daily living skills, much less do his favorite activities. Helping his mother make cookies will become a distant memory. If there is a treatment out there that can help children with this diagnosis, slow the course, and give them a chance to live longer, fuller lives it needs to be approved and utilized. The drug, [drug name], cannot help my son specifically. But it should be utilized to help as many boys (and girls) as it possibly can benefit to slow the progression of this disease and give them a chance. In addition, follow along exon skipping therapies need to be given swift trials and approval. My son could specifically be helped and given more time to live with a skip of exon 53. Our children don't have the time for placebo trials. Natural history of this disease has already been studied. As a parent, I am willing to accept risks that could come with these treatments. While my son and a large amount of the Duchenne population can be helped with exon skipping

therapies, there are others our with other genetic abnormalities causing DMD such as duplications, splices, and point mutations. We cannot ignore these boys (and girls) either. Treatments for all are needed.

### **Two Sons, Twice the Heartache**

While many of my son's middle school friends are just rolling in from practice, its a typical night at my house. Bed time starts no later than 7:00 for my two sons with DMD. Kevin is 13 and Kyle is 9. Their friends grab a snack or start their homework but my boys aren't strong enough to get drinks out of the fridge or pick up their own backpacks. Just like their friends, our home is full of laughter (boys will be boys) but between the requests for help to get on the toilet, off the toilet, showering, dressing, picking up anything that might have fallen to the ground, stretching 2 sets of tight heel cords and quad muscles then putting on leg splints, and hooking up BiPap machines, there isn't much time for goofing around. I need to keep them focused because this bedtime routine is long enough. Bedtime wraps up about 9:30 with helping the boys get positioned in their beds. It never dawned on me that they would get to the point where not only would they need help getting comfortable in bed, but also would call me every time they needed to roll over during the night. It is so tiring for me, but who am I kidding, my sons exhaust themselves every time they walk to the bathroom. Last week as I helped, Kevin, my 13 year old son get into bed he moved his arm to hold my hand and told me its getting harder to use his arms now. He asked me, "Someday is it going to get too hard for me to wave to my friends?" I told him probably. He cried and I held his hand and I hugged him. No mother should have to comfort her son like that. I know in a short while I will be going through this same scenario with his little brother. No mother should ever have to do it twice. Kevin has said he would do anything to find a cure for DMD and I believe him. He prays every night at dinner for a cure. He and his brother don't even flinch as they endure blood draws, MRIs, and skin biopsies as part of clinical trials. These boys are brave but they get are getting tired. They are too young to be getting tired. They are truly heroes.

### **Back of the Bus**

Duchenne Muscular Dystrophy, you have rocked our world! We were given this beautiful baby boy to love, cherish, and enjoy him for many years. Instead, you



**KYLE**, 9 years old; **KEVIN**, 13 years old

have taken over our wonderful young man. You did not even hold out for a little while. From birth you were there interrupting his life. You did not allow him to run, jump, skip, nor all the other fun things a child should enjoy. How do you answer a child when he asks the questions: "Why" "Will I ever" "How bad, is bad enough" "What will I do when I am older?" As the parent, you know the answers are not good at all. Then add all the extra things in there that are difficult for a teenage boy to comprehend, such as, taking him to the bathroom, getting him dressed, giving him baths, brushing his teeth, rolling him over every couple of hours during the night, etc. Some of the worst symptoms for us are: contractions, heart problems, seizures, pressure sores, and many more. All of the symptoms just lead to more problems such as constipation, numbness in feet and legs, scoliosis, loss of strength including neck. Imagine your 14 year old being held like a newborn because he does not have the neck muscles to support his head. As for therapies, WE the families do more than any outside source. We fight for our son's to live, move, breathe, eat, sleep, and to live normal lives on a daily basis. We need your help! We want a cure! Our boys deserve more than prednisone! They deserve the opportunity to get up and move! We need more trials for all different stages of DMD. Our son has NEVER qualified. It appears that when the child has severe DMD he is written off as a lose, lose situation. He is



**JORDAN**, 14 years old

our win, win! He lights up our world. We want him with us for many years to come! Please help!!

### **Progression of Duchenne**

It can happen to anyone. Duchenne knows no borders of geography or ethnicity. My beautiful boy had trouble walking, getting off the floor. He never crawled. Doctors assured me “He’ll grow out of it.” My son’s physical abilities peaked at 7, but he frequently fell even at a young age. His body could never keep up with his dreams and desires. It was with bittersweet relief when he broke his ankle at 10 and stopped walking. That’s right. I WANTED my boy to use a wheelchair. NO MORE FALLING. Whenever I wish for life without a wheelchair, I simply look back to when my child would spontaneously crumple to the ground, unable to catch himself and slam his face onto the pavement, carpet, or his own feet. The horror of not being fast enough to catch your child, and then living your sons agony with him as he cries out in frustration that his body WON’T DO WHAT HE NEEDS IT TO. While I live in this “honeymoon phase” of my son’s relatively good health, I’m constantly aware that his heart may give, he may not recover from a simple flu. The likely hood of dying young stands in stark contrast to my son’s lust for life and adventure. Dying happens to us all, there’s no denying that. But within that reality, if given a choice for my son? It would not

be drowning in his own fluids. It would not be from his heart giving out as a young man. It would be after meeting his grandchildren and living a long, happy life. A life where he could choose independence from his parents, and where his body doesn’t give out from lack of vitality, but from many, many years of use. We struggle with strength. As my son grows, his weight makes it difficult for transfers... to the toilet, to the bed, to an airplane seat for travel. He has so many dreams to go places, and I agonize over the day when his doctors tell us he must have rods fused to his spine to maintain his breathing capacity. With that operation will go his ability to hop on a plane and visit family. With each procedure he is likely to become more isolated from his peers. My social boy who loves to interact with people of all ages becomes more and more alone. My son’s type of Duchenne is rare. There can be no hope for him if a drug found safe and effective and benefits a large portion of the Duchenne community cannot be approved. Blocking the progress of this drug’s availability will dissuade researchers from continuing the search to help all Duchenne patients. It is maddening that after 50 years there is finally an effective medicine that helps stave off the inevitable decline of this vicious disease for a decent percentage of those effected, and it’s progress is being stonewalled. It is simply unacceptable. It is unconscionable.

### **Exon Skipping as a Class**

Dear FDA, Our son was diagnosed just 5 years ago at age 9. We consider our family to be lucky because in the past 5 years Duchenne has only taken away our son's walking, standing, dressing and bathroom independence. So far we have managed these things. We are also lucky because in the past 5 years we've only had to purchase (out of pocket), one manual chair for travel, one power chair, one ceiling mounted lift, one freestanding lift for travel, one mini-van with turning seat and lift, one mini-van with manual ramp and lockdown element, one-one story house with wide doorways and halls, a couple of doorway ramps and one suitcase ramp for travel. Not sure how, but we have managed these things as well. We are also lucky to be fighting Duchenne in the past 5 years since there are many excellent scientists working internationally to find ways to treat everyone with Duchenne. We are doing our part to make things work for all our sons. Now it's your turn. Approve Exon Skipping as a "class" instead of making each and every exon skipping compound go through years and years of individual clinical trials. Approving exon skipping as a "class" may not be risk free but under the circumstances it is still the right thing to do. Do the right thing so we can have a realistic hope that our son(s) will live long enough to see regenerative medicine. Do the right thing so they will have the chance to stand, walk, dress or bathroom independently. Do the right thing because if you were in my shoes, you'd want the same thing for your own son. They deserve nothing less. Thank You.

### **Need to Look at Duchenne Differently than Other Diseases**

I want the FDA to know that as a mother with a child with Duchenne, we are desperate for any treatment that can help our son to be stronger or live longer. We have a different requirement for safety and trials of the medications/treatments. This is not a regular disease. Drugs should always be fast tracked. There needs to be a different set of requirements. This is imperative for our family's well-being.

### **Three Sons with Duchenne in One Family**

My name is Betty Vertin. I live in Hastings, NE with my husband and five children. We have a 12 year old daughter Lexi and 4 sons; they are Max, 8, Chance, 6, Rowen, 5, and Charlie, 3. In July of 2010, when I was just 10 weeks pregnant with our youngest, our

son Max was diagnosed with Duchenne. In a matter of minutes a specialist told us what no parents ever want to hear: that our son would be wheelchair bound by 9, need our help with activities of daily living as a teenager, and would die before his 20th birthday. We woke up that morning with a beautiful healthy son and went to bed that night grieving the loss of a son that would come too soon. In the course of the next year, Rowen and Charlie were given the same fatal diagnosis. We believe, or at least hope the progression of the disease may not happen as quickly as the first doctor described to us; but we do know there is no cure and our sons will die because of the disease if there is no cure or at least better treatment options. I am sharing with you because three of our four sons have Duchenne Muscular Dystrophy. I am sharing with you because I am scared that 3 of my 5 children could die without a cure or better treatments of Duchenne Muscular Dystrophy. My sons are young; but still face daily challenges because of Duchenne Muscular Dystrophy. Max is our oldest son, he is 8. Max's muscle strength is in a decline. There are things he could do three or four months ago that are becoming increasing more difficult for him. He is falling more often. Two weeks ago he fell and hurt the tendons around his knee. He limps and uses his medical stroller more now than ever before. We hope that as his tendons heal, his mobility will come back. However, because of Duchenne Muscular Dystrophy there is no guarantee that he will bounce back. Just yesterday he fell trying to play soccer, hit his head and has a concussion. He was falling like this three months ago. The hardest for me to swallow is that Max does not always finish putting a lego set together. That was unheard of a couple of months ago; Max would sit for hours to complete a set, but now he doesn't seem to have the stamina. As a parent this is very hard to watch, no one should watch their children regress. Parents dream of watching their sons grow into men, watching their dreams come true. Duchenne and the lack of treatment threatens to steal this dream from us. It threatens not just the loss of one son, but three sons. It threatens to take half of my children from me. My husband and I feel we are proactive, we are doing everything within our power to save our children. We make financial sacrifice to take our children to the best doctor available (14 hours from our home), we follow a strict diet, we home stretch, we fund-raise to help finance research. In fact, Max participates in the [clinical research] study and [drug] trial in hopes



**MAX**, 8 years old; **ROWEN**, 5 years old; **CHARLIE**, 3 years old

that not only we help, but that he can have access to a drug that may save him and eventually his brothers. Our experience with the [drug] trial has been heart wrenching and I would like to share with you why. We screened Max for the study 6 months ago, before his decline, and he did not make it into the study because he walked too far during the 6 minute walk test. We re-screened this February and he made it in, and although we are happy he did, it was because his muscle strength had declined and he was no longer able to walk too far. If there had been no 6 minute walk test, Max could have had access to [drug] before his decline and his strength could have been saved. We do not know if Max is receiving a placebo or the actual drug. However, based on the changes we have seen in his strength the last couple of months, I think it is very likely he is on the placebo. If there was no placebo, his change in muscle strength, his decline could be stopped. This is why I believe with all my heart, that every day that it takes to approve a drug, is a day my sons do not have. It is why I believe so strongly is a faster approach to approve the drugs that could help. I want my sons to be saved. I want to watch all 5 of my children blossom into amazing adults and see all the things they accomplish. The possibility

that I might see only two children live, should not be a possibility I have to consider as I think of the future. I have attached a picture of our family so that you can see those cute boys and know what we are fighting for and that you would join our fight to save them. Max, Rowen, and Charlie are the three we are holding. Sincerely, A mother in love with her sons and not willing to let them go. Betty Vertin

### Hope for the Future

I want the FDA to know that when Duchenne enters your life, there is no "son, you can grow up and be anything you want to be" discussion anymore. I want them to know that it changes everything about parenting you ever thought you knew to be true. Before Duchenne, I pushed my children to do their best at school, and set goals for themselves. Give 100%, isn't that what we're supposed to do? But just how important is education for a child that may not live long enough to utilize it?? How important is discipline when by the time my son reaches the age where he can start to actually reflect on and practice our teachings to him, he will be struggling just to breathe and eat?? I made a commitment to him when he was diagnosed - I will knock down every door. I will

raise as much money as I can. I will search the world over for any opportunity to save him. I will give up my career in order to focus on this goal full-time. I will give up every single personal goal I have for myself so that he can have what he needs/wants from me. After all, God chose me to be his mother for a reason, and I understand that he is a gift for which I will always be grateful. I want the FDA to consider the possibility of not only acting as a regulator, but as a facilitator. A partner. While we recognize the need for safety and appreciate their efforts, our fight is to save our sons' lives. NOW. I would love to see the FDA put a panel together that interacts with our community about clinical trial design. It was apparent from last year's PPMD Conference there is a disconnect between regulator and clinician regarding endpoints. The FDA says "we review what we are given" and the clinician says "we want to know what is an acceptable endpoint and when we ask we get no information"...why can't there be more of a partnership here? The FDA can be our true heroes here. No other country has approved a treatment for this God awful disease yet. Our country can show true leadership and compassion by accelerating the process for approval and partnering with us to get this done.

### Message of Urgency

As a parent, I think it was the hardest question I've had to answer (yet): "Will I never be able to get up on a stool again without help?" Automatically my rehearsed optimism kicked into gear. "We hope that's not the case. We hope that all the doctors working on new medicines will find a cure." My son was 3 years old when he was diagnosed with Duchenne muscular dystrophy. But, we dared to believe, this was a new era for the disease. Medical science had had so many recent break-throughs. A cure was just around the corner. Seven years later, my son is getting a new set of wheels -- a wheelchair, that is -- for his 11th birthday. Still no cure. The studies go on, but still no real break-throughs -- at least not for my boy, who can't go trick-or-treating without his dad to lift him up the front steps to ring the doorbell. We push him to do well in school, not knowing if he will be able to hold a pencil by the time other kids his age learn to drive. Most kids learn independence as they grow. Mine gradually relinquishes every milestone he gains. God can't help my son. But medical science can. Frighteningly, Duchenne moves faster than new drugs become available. If you have it in your power, if there



**JACKSON**, 7 years old

is anything you can do to speed the process, to make hope and faith mean more than empty words, please, please do so. For me, for my son, the time is now. Down the road is too far for him to walk. His chance is here and now. Let it be true that the doctors can help. Let it be true that a stepstool will be only the beginning of the heights he will yet achieve.

### Harmonizing between Regulatory Bodies

I want them to know that my son is a beautiful, seemingly healthy boy that is dying inside. His muscles aren't and can't grow until they allow us medicines that can slow the progression of Duchenne. I understand the need for caution and care, but I also know that while drug companies are allowed to give medicines in other countries, our children are dying. We should be able to use studies and data from other countries without having to jump through duplicative hoops in America just to say we did! Parents should be able to decide the risk/benefit of a drug that has gone through and passed preliminary testing. I would rather my son die trying and fighting than waiting and wondering and wishing. Please help these young men and women who are RELYING on YOU for help. Please don't slow down the progression of drug development. Please find ways to speed it up! I am one parent willing to take an educated risk! Thanks!

These are only a small fraction of the stories. We encourage you to visit [ParentProjectMD.org/MyVoice](https://ParentProjectMD.org/MyVoice) to hear more from our community.



# CONCLUSIONS

As a final message, several participants **thanked the FDA for their commitment** to drug development for rare disorders and for Duchenne more specifically.

*“We understand all that the FDA does for patients. In addition, we especially appreciate that you listen to our community and are trying to rethink your expectations for trials with rare diseases. Thank you for all you do!”*

Families of individuals with Duchenne muscular dystrophy would like the FDA to feel strong pressure to move as quickly as possible, because of how Duchenne muscular dystrophy progresses. Delays mean that people with Duchenne lose abilities and possibly their lives. They hope for increased flexibility in the design of clinical trials, and higher tolerance for risk from the FDA—important steps in their quest to give themselves/their children a chance for a better future.

For more information please contact Holly Peay or Ryan Fischer.



# ABOUT PARENT PROJECT MUSCULAR DYSTROPHY

Duchenne is a fatal genetic disorder that slowly robs young men of their muscle strength. Parent Project Muscular Dystrophy (PPMD) is the largest most comprehensive nonprofit organization in the United States focused on finding a cure for Duchenne muscular dystrophy—our mission is to end Duchenne.

We invest deeply in treatments for this generation of young men affected by Duchenne and in research that will benefit future generations. We advocate in Washington, DC, and have secured hundreds of millions of dollars in funding. We demand optimal care, and we strengthen, unite and educate the global Duchenne community.

Everything we do—and everything we have done since our founding in 1994—helps boys with Duchenne live longer, stronger lives. We will not rest until every young man has a treatment to end Duchenne. Go to **[ParentProjectMD.org](https://ParentProjectMD.org)** for more information or to learn how you can support our efforts and help families affected by Duchenne.

# Parent Project Muscular Dystrophy

LEADING THE FIGHT TO END DUCHENNE

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