Chapter 13
U.S. Patient Advocacy Groups

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For newly diagnosed muscular dystrophy (MD) patients and their caregivers in the U.S., the first point of call for information and support should be the Muscular Dystrophy Association (MDA, www.mda.org). In addition, there are disease-specific groups for several of the nine forms of MD, as shown in Table 13.1.

All Forms of MD

The MDA is the world's leading nonprofit health agency dedicated to finding treatments and cures for MD, amyotrophic lateral sclerosis (ALS), and other neuromuscular diseases. The Association does this by funding worldwide research; by providing comprehensive healthcare services and support to MDA families nationwide; and by rallying communities to fight back through advocacy, fundraising, and local engagement.

The organization is funded by individual private contributions and cooperating organizations, providing research, services, and education. Currently, over 250 research projects are funded through the MDA. Although a national group, the MDA makes its resources and information accessible through local chapters, with 200 clinics throughout the states and 100 local offices. Some 44 of those clinics are MDA/ALS centers. MDA holds annual biannual clinical and scientific conferences where cutting-edge research and clinical trial information is presented. In addition, around 3,800 children attend MDA summer camp every year, and MDA's online Transitions Center is a clearinghouse for resources to support young adults seeking employment, education, independent living, and community involvement; blogs are posted twice weekly from young adults sharing their experiences living with neuromuscular disease.

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Table 13.1 Patient advocacy groups for MD

Type of MD	Sources of information and support
Duchenne MD	MDA (MDA, mda.org)
	Parent Project Muscular Dystrophy (PPMD, http://www.parentprojectmd.org)
	The Foundation to Eradicate Duchenne (http://duchennemd.org)
	Duchenne Alliance (http://www.duchennealliance.org)
	Other organizations around the world are listed at http://www.parentprojectmd.org/site/PageServer?pagename=Connect_partners, http://www.treat-nmd.eu/dmd/patient-organizations/, https://www.duchenneconnect.org, & http://www.cureduchenne.org
Becker MD	MDA (http://mda.org/disease/becker-muscular-dystrophy)
Congenital MD	MDA (MDA, mda.org)
	Cure CMD (http://curecmd.org)
Distal MD	MDA (http://mda.org/disease/distal-muscular-dystrophy/types)
Emery-Dreifuss MD	MDA (http://www.mda.org/disease/emery-dreifuss-muscular-dystrophymda.org)
	National Organization for Rare Disorders (NORD, https://www.rarediseases.org)
Facioscapulohumeral MD (FSHD)	FSH Society (http://www.fshsociety.org)
	MDA (MDA, mda.org)
	PNW Friends of FSH Research (http://www.fshfriends.org)
Limb-girdle MD	MDA (MDA, mda.org)
	Jain Foundation (http://www.jain-foundation.org)
Myotonic dystrophy (DM)	MDA (MDA, mda.org)
	Myotonic Dystrophy Foundation (http://www.myotonic.org)
Oculopharyngeal MD (OPMD)	MDA (http://mda.org/disease/oculopharyngeal-muscular-dystrophy)
	NORD (https://www.rarediseases.org/rare-disease-information/rare-diseases/byID/1182/viewAbstract)

MDA also hosts 150 support groups across the country. MDA's advocacy program, based in Washington, DC, seeks to make the MD community's voice heard and to expand resources for those with neuromuscular disease. By informing and educating legislators, MDA aims to accelerate development of new therapies. Individuals can sign up to be an MDA advocate and receive regular advocacy updates on the organization's web site.

The MDA publishes a quarterly magazine, *Quest Magazine*, which is sent free of charge to families registered with MDA and is also archived online at http://quest.mda.org. The organization also has an online-only *MDA/ALS Newsmagazine* at http://alsn.mda.org. MDA is also active through social media channels, including Facebook and Twitter, enabling patients and caregivers to connect with one another and providing the latest research updates.

Duchenne MD

Parent Project Muscular Dystrophy (PPMD) focuses on finding an end to Duchenne MD specifically. In total, this group has invested over $45 million dollars in research, which has leveraged over $500 million in additional funding. PPMD has been involved in several steps forward for the MD community, including providing the FDA with the first-ever patient-initiated guidance to help accelerate development and review of potential therapies for Duchenne MD. PPMD also recently announced that a gene therapy study that it had funded might improve walking ability in MD. Historically, the group was instrumental in the passing of the MD Care Act in 2001. There are also a number of projects listed on their web site that need funding and lists of projects they have supported financially.

The PPMD web site has a helpful section, labeled as "Connect," which provides links for the group's monthly e-newsletter, updates on upcoming events, current research, and other MD groups. The organization has a mobile application for iPhone and Android users that informs patients of new clinical trials and other news, as well as location and contact information for nearby clinics. The PPMD Facebook and Twitter pages are regularly updated with pictures and news from the MD community, both scientific and patient-related. PPMD has also created a community page where visitors can post a profile in order to meet other members, read blogs, and share pictures. Many people who are affected by MD find that attending an event, having a chance to meet other patients or family members facing similar challenges can bring a sense of comfort and connectedness. The PPMD holds annual Connect Conferences and Meetings, typically attended by around 500 families.

There are other organizations keeping the end to Duchenne as a priority. CureDuchenne states on its website that it is a "national nonprofit that raises awareness and funds to find a cure for Duchenne muscular dystrophy." This organization has helped fund two companies (Prosensa and Sarepta) that are seeking FDA approval for drugs for MD. CureDuchenne's research investments have leveraged around $100 million from government agencies and pharmaceutical companies. In 2014, they hosted the sixth Climb for Duchenne, where "teams of people across the country can climb various mountains, hills, or tall buildings" to raise awareness and funds for CureDuchenne. The site encourages advocacy for fighting Duchenne by creating links for starting individual fundraisers.

The Foundation to Eradicate Duchenne (FED) was created in 2002 by Dana and Joel Wood, residing in Virginia, after their son was diagnosed with the disease. The Foundation's web site says it has worked with others to achieve millions of dollars in federal earmarks for Duchenne MD research and a significant increase in the attention devoted to DMD at the National Institutes of Health. Additionally, through the FED and other fundraising efforts, the group has raised nearly $10 million in private donations and worked with Congress to secure nearly $40 million in federal appropriations. While not updated frequently, this group's social network pages include several links for contact information from staff and for general questions, advocacy, and donations.

The Duchenne Alliance is an "alliance of independent non-profit organizations," with various academic and industry research projects inviting donations. The group also has plentiful blog entries and is active on Facebook and Twitter.

While the majority of patient advocacy resources focus on Duchenne, there are also resources for other types.

Becker MD

Due to its mechanistic similarity to Duchenne MD, Becker MD is covered by many of the same groups that focus on Duchenne, particularly the MDA.

Congenital MD

Cure CMD provides extensive resources for patients with congenital MD. The group has a mission to bring research, treatments, and in the future, a cure for CMD, by working globally together with dedicated parent, government, and research advocates. By focusing on this mission, Cure CMD aims to find and fund high potential research and clinical trials. MDA also provides extensive clinical and research funding, and other support services to individuals with CMD.

Distal MD

The MDA supports patients with this type of MD.

Emery-Dreifuss MD

This type of MD is also supported by the MDA, and by the National Organization for Rare Disorders, which provides links to various other resources at http://www.rarediseases.org/rare-disease-information/rare-diseases/byID/590/viewAbstract.

Facioscapulohumeral MD

The FSH Society (Facioscapulohumeral Society) is a world leader in combating FSHD. The Society's purpose is to conduct research, increase awareness, understanding, and education on FSHD. This is especially important as FSHD may be one of the most common adult MDs, affecting men, women, and children worldwide.

MDA has also spent millions on FSH research and support services for families, and the majority of individuals are seen in MDA clinics.

The FSH Society has provided seed funds and grants to pioneering FSHD research areas and education worldwide and created an international collaborative network of patients and researchers to support research relevant to understanding the molecular genetics and causes of FSHD. The FSH Society provides strategy for FSHD research, therapeutics, and clinical trials readiness, recruiting qualified researchers and clinician-researchers, selecting research proposals, evaluating research proposals, granting fellowships, and monitoring ongoing projects and research opportunities. Grant making to FSHD researchers and clinicians is one of the largest components of the FSH Society and these efforts have led to more than 300 publications acknowledging this support in scientific journals.

Recent advances in understanding the molecular genetics and cellular biology of FSHD have led to the identification of potential therapeutic targets. The Society's main focus is to gather more support for research from the U.S. government by submitting both oral and written testimonies to Congress, all of which can be read through links on the site.

Meetings, symposia, workshops, and networking activities are one of the most successful programmatic components of the FSH Society. Through the FSH Society staff and its web site portal at www.fshsociety.org, Facebook page, Twitter account, Yahoo! Groups bulletin board, e-mail ListServ, and quarterly newsletter the *FSH Watch*, FSHD patients have found ways to be useful to one another and to basic and clinical researchers working on their disease. This group's web site is extremely abundant with information, much appreciated by the author, who is an FSHD patient herself. The support patients receive from one another through sharing their common experience is invaluable and immeasurable. The FSH Society acts as a clearinghouse for information on the FSHD disorder and on potential drugs and devices designed to alleviate the effects of the disease. It fosters communication among FSHD patients, their families and caregivers, charitable organizations, government agencies, industry, scientific researchers, and academic institutions.

The FSH Society also provides dedicated support, education, and outreach services to patients, professionals, researchers, and families in need of assistance. The Society responds to inquiries by phone, web, and e-mail from newly diagnosed patients, patients, family members and spouses of FSHD patients, and professionals.

The FSH Society helped educate and recruit patients into research studies to help facilitate the production of the world's largest resource for FSHD biomaterials that are being made available to researchers worldwide. The Society hopes that this strategy will help with better reproduction, validation, and corroboration of research results by providing the community with a high quality and high number of well-controlled FSHD cell lines that multiple research groups can independently access. Publications, literature, education, patient support, social networking, and research networking combined are the most significant components of the FSH Society.

Limb-Girdle MD

There are many foundations that focus on individual subtypes of limb girdle muscular dystrophies (LGMDs), both in the U.S. and abroad. These foundations are often started by families of individuals with the disease and usually focus both on patient advocacy and on supporting research towards finding treatments and cures. MDA has also invested a lot of funding for research and support for families with LGMD.

Although LGMDs share common symptoms, the diseases are caused by mutations in a large number of genes. This large genetic diversity, the high cost of genetic analysis, and the refusal of some health insurance companies to cover genetic diagnosis make it difficult for individuals to obtain a definitive diagnosis. To address this problem, a consortium of LGMD family foundations was formed in 2014 and includes the Cecil B Day Family, Inc (LGMD2B), Coalition to Cure Calpain 3 (LGMD2A), Jain Foundation (LGMD2B), Kurt+Peter Foundation (LGMD2C), LGMD2D Foundation, LGMD2I Fund, and McColl-Lockwood Laboratory (LGMD2I).

Led by the Jain Foundation, the LGMD consortium created a new diagnostic program (http://lgmd-diagnosis.org) that offers free genetic sequencing to individuals with unexplained muscle weakness. Patients can apply for the program by taking a short quiz or their physicians can apply on their behalf using the Jain Foundation's Automated LGMD Diagnostic Assistant (ALDA—http://www.jain-foundation.org/alda) to determine eligibility. Qualified patients send in a saliva sample and receive a genetic report that includes results from a gene panel of 35 genes known to be involved in various forms of LGMD as well as other muscle diseases with similar symptoms. The diagnosis program launched in late 2014 and is already succeeding in its goal of identifying a large number of individuals with LGMDs.

Many of the LGMD consortium members focus on patient advocacy and have patient registries including the Jain Foundation, Coalition to Cure Calpain 3, LGMD2D Foundation LGMD2I Fund, and the Kurt+Peter Foundation. The identification of a large number of patients with the LGMDs studied by the foundations will help each foundation in their goals of curing each disease.

The Jain Foundation (http://jain-foundation.org) has a mission of curing muscular dystrophies caused by mutations in the dysferlin gene, which includes the clinical presentations Limb-Girdle MD type 2B (LGMD2B) and Miyoshi muscular dystrophy 1 (MMD1). The foundation is privately funded and does not solicit funding from patients or other sources. Its strategy includes funding and actively monitoring the progress of scientific research projects in key pathways towards a cure, providing financial and logistical support to promising drug candidates to accelerate them to clinical trials, funding clinical trials and studies, encouraging collaboration among scientists, and educating LGMD2B/Miyoshi patients about their disease and helping them with their diagnosis.

Coalition to Cure Calpain 3 (C3) (http://www.curecalpain3.org) was founded in 2010 for the specific purpose of funding research efforts focused on understanding the biology of and finding a cure for LGMD2A. This disease is also sometimes

referred to as calpainopathy because it is caused by mutations in the calpain 3 gene. The organization was created by people with LGMD2A for people with LGMD2A, as both founders have the disease. The main focus of C3 is on supporting researchers and encouraging collaboration among scientists rather than on providing services to those who have the disease.

The LGMD2D Foundation (http://lgmd2d.org) is a non-profit private foundation whose mission is to expedite the development of a cure or therapy for Limb Girdle Muscular Dystrophy 2D (LGMD2D), which is caused by mutations in the alpha sarcoglycan protein. In addition to educating patients and physicians, the LGMD2D Foundation maintains a patient registry, funds and monitors research and progress, provides financial support to accelerate clinical trials, and encourages scientific collaboration.

The LGMD2I Research Fund (http://www.lgmd2ifund.org) is a not-for-profit focused on expediting the development of a treatment or cure for Limb Girdle Muscular Dystrophy 2I (LGMD2I), which is caused by mutations in the fukutin-related protein. The foundation does this by building a comprehensive view of the entire LGMD2I research landscape, supporting the most promising research projects, and coordinating and managing the scientific process.

The Kurt+Peter Foundation (http://kurtpeterfoundation.org) was formed by the family and friends of Kurt and Peter Frewing to raise money and direct it into the hands of the researchers who have the best shot at developing a treatment or cure for LGMD2C, which is caused by mutations in the gamma sarcoglycan gene. Since 2010, the Kurt+Peter Foundation has raised more than $1 million for research into LGMD2C. Among other initiatives, the foundation is currently funding development of an exon skipping compound that the foundation hopes will treat the majority of LGMD2C mutations.

Myotonic Dystrophy (DM)

The Myotonic Dystrophy Foundation is focused on supporting and driving the research, resources, and community capacity needed to achieve the first clinical trials for myotonic dystrophy treatments, while providing comprehensive support and education to families living with this disease. The foundation's web site provides listings for support groups, a blog, details of ongoing research, and links to social media sites such as Facebook and Twitter. Both are updated regularly, and the organization is partnered with MDA. Individuals may join the Myotonic Dystrophy Family Registry, which gives the members access to research data and anonymous information regarding individuals also living with this particular genre of dystrophy.

MDA also has a focus on DM.

Oculopharyngeal MD (OPMD)

This form of MD is covered by the MDA and NORD.

Outside of the patient advocacy groups, other sources of information on MD include medical, healthcare system, academic, and government institute web sites. Examples include WebMD, Medline Plus, Medscape, Genetics Home Reference, the Mayo Clinic, Johns Hopkins Medicine, the University of Maryland Medical Center, National Center for Biotechnology Information, and National Human Genome Research Institute.

Overall, patient advocacy is extremely important in encouraging new research in every kind of MD, increasing awareness of the disease among policy-makers, and providing resources to positively impact the everyday life of patients living with MD. This can be a rewarding area for patients who wish to be involved in changing their own lives and those of others afflicted by this group of diseases.

Non-U.S. Patient Advocacy Groups

Because some of the forms of MD are so rare, little information is available in the U.S.; therefore, a patient with a very rare form of MD may need to look outside of the U.S. to get additional information. Although beyond the scope of this chapter to discuss in detail, one Website worth mentioning is Treat-NMD. Treat-NMD (www.treat-nmd.eu) is a neuromuscular network that provides a list of global registries that can be accessed at http://www.treat-nmd.eu/resources/patient-registries/list/. Queries can be made by going to the disease information tab, picking a disease, and then on left-hand side menu, a "patient organizations" tab can be clicked to view the worldwide list of organizations for that disease.

Learning About FSHD: Tips for Patients

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As a MD sufferer—I was diagnosed with FSHD at Duke University's MDA Center in 2003—the best advice I could offer someone who is newly diagnosed would be, "Don't hesitate to educate yourself on your disease." Figuring out what exactly you are dealing with and how you can help yourself and your loved ones will make the situation easier to cope with as a whole.

Connect with Advocacy Groups & Other Patients

Fortunately, awareness of MD continues to grow. Scientists continue to produce more findings and hopefully one day, there will be a cure for every type

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of MD. As our world becomes more connected, it is easier to read up on updates in research, learn the symptoms and causes of your malady, and connect with others through social media. Patient advocacy is vital in fighting MD, as it is with any medical condition, especially those with limited awareness and no cure at this point in time. There is always the option of making a donation to organizations that fight against MD, setting up a fundraiser for the cause, or working at a summer camp for children afflicted. Using your voice is an important tool as well. The MDA provides a page in which you can find your elected officials who vote on important pieces of legislation affecting MD patients and their families.

Ask for Help When You Need It

As a patient, I understand that this disease comes with more than just physical side effects. It can be humiliating, frightening, stressful, disheartening, and confusing. I was diagnosed with depression in high school after I began to accept the changes going on in my body, and when I began to try and accept and recognize my limitations. Even without the daily struggle of coping with MD, it can sometimes be difficult and embarrassing to admit our weaknesses and to ask for help when we need it. We want to be independent, we want to take care of things ourselves, we want to say "I did this for myself. I didn't need help."

I am still learning how to ask for help. I am learning how to undermine my stubbornness, I am learning to talk about and admit openly the simple truth that I am physically weaker than most people that I encounter. I am learning to offer a compromise when invited to do things I may not have the strength to do, or learn how to tell others I'm going to have to "sit this one out," I am learning to watch others run and dance and climb with joy instead of resentment, jealousy, and anger.

Try Exploring New Pastimes

It is easy to feel cheated when you don't have the same opportunities and it is easy to feel excluded. I began dance when I was three years old. I fell in love with it. It was a way to be active in a fashion that I felt coincided with my very soul, and it was a way for me to get stress out. I took tap dance classes, I took ballet, I took hip-hop, modern/contemporary. Being in a studio was like being at a different kind of home. When I was forced to take a lower level dance class as a sophomore in high school, the same one I had taken as a freshman, I was angry. I couldn't physically keep up with the higher level classes, and it tore at me. I had been a dancer for years and years, I could choreograph a routine in a short amount of time, I knew how to do all the moves, I heard counts and beats in every song I heard. I day-dreamed routines in my mind, I couldn't listen to a song without wanting to move some part of me.

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After my sophomore year, I admitted defeat to myself. I stopped dance altogether. I canceled my subscription to my dance magazine and shoved my tights and leotards in the bottom of my drawer. To give up something that seems like your life, something that you're passionate about, is torturous. It brings about some of the emotions I previously mentioned. As someone with MD, you are most likely able to relate. To give up something like that, and on top of that, sometimes even simple daily tasks, is a complete life changer. And seeing others accomplish things you wish to as well is frustrating.

Enjoy Simple Pleasures

There is, however, good news. There is always a silver lining if you choose to look close enough. You did not choose this, your loved ones who suffer from MD did not chose this. Blaming yourself, blaming others, and being angry is something that will cripple you even more. Let any anger you feel serve as motivation for something great, or throw it away. You may be unable to run down the soccer field, you may not be able to climb mountains by yourself, you may have to give up things you find hard to. There are other things you can try, other hobbies you can find. I threw myself into art and writing, and found that I have somewhat of a talent for both that I am working on furthering developing. Negative emotions are hurtful, but you can put them into words that others may relate to, you can let them flow through a paintbrush, you can sing them for a loved one. There is still a beautiful life that you can fit into just as easily as anyone else, and you are no lower than anyone else just because maybe you need help reaching for that cup on the top shelf. You have a unique perspective as a person with MD. You may possess a greater appreciation of the simpler things in life, you may be less judgmental as you understand that everyone has their own struggles in life, and just because you can't always see or understand them does not mean they do not exist. You may be more compassionate due to the fact the compassion towards yourself is greatly appreciated, that when someone asks for help it might mean the world to them just as my friends piggy-backing me up hills without annoyance or frustration means the world to me. You may learn how to cope better, or you may develop better coping skills from having to deal with so much yourself on a daily basis.

I struggled many years worried about what I later discovered were rather silly things. People who truly love you DO want to help you, even if they don't always know the right way, the right things to say or do. While I was generally embarrassed and felt sometimes annoying, I've been told many times things like "It's not even a big deal. It's cute anyway! I'd give you piggy-back rides regardless if you wanted."

It takes courage to be open. Walking in public places might get you many stares or whispers. I've heard countless mentions of how thin I am, or how my

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gait is slightly off. While it is sometimes hurtful, I've learned to either address the situation by using it as an opportunity to politely educate someone, or simply ignore it. What others say of you says more about them than it does you. Some people simply haven't heard of MD. It doesn't necessarily mean they are uncaring or cruel. A girl I once sat by in a class of mine used to tell me nearly every day that I should eat more, because I looked sickly. For several weeks I either laughed it off or mumbled back things like "yeah, maybe." After a while I finally mustered the courage to tell her that I was only thin because of my condition, and I actually ate more than a man going through college. Not only did she feel extremely guilty (which wasn't my goal), but she learned something new and started helping me gather my things after class, offering to carry my books if I needed.

Smile the Best Way You Can

Never take situations like this personally; you are beautiful no matter your capabilities or your appearance. Living with any disease is hard. MD has posed many obstacles for me, has brought many tears, and has made me question things I probably normally wouldn't without it. I am growing a greater appreciation for MD every day, however odd that may sound. It has taught me compassion, it has taught me forgiveness, it has taught me humility, courage, and appreciation. Life does not often go as planned, and we must learn to accept that and use it to our advantage. So, while people telling to smile bigger has always bugged me due to the fact I've lost some muscles in my cheeks, I urge you to smile in the best way you can. It is okay to hurt, it is okay to cry, it is okay to feel sad and lazy some days. It will rain some days, just remember the good weather, and know that it is coming. Don't be afraid to let others help you, and don't be afraid to offer your help to others. I wish you much peace, plenty of love, and safety in your journey.