Becker Muscular Dystrophy (BMD)

Processing a new diagnosis

A diagnosis of BMD can be a huge shock for the person with the condition, parents, siblings, extended family members and friends. It is normal to feel an overwhelming mix of grief, confusion, anxiety, loneliness and helplessness as your and your family's life has been forever changed. Everyone will have different ways of getting through this time but it's important to know that help and support are available for you, your child and your family. Hear advice on processing your diagnosis from a community member with shared experience.

The video is available at **https://youtu.be/A9XM71knDVw.**

Getting help processing a new diagnosis

There are many ways you can seek support as you process a new diagnosis:

- A psychologist is a university-qualified health professional who can help you talk about your thoughts and feelings to understand and cope with the challenges you and your family are facing. Visit our page on **Psychology** for more information.
- A counsellor is a trained professional who can help you talk about and work through problems. Visit our page on **Counselling** for more information.
- A social worker can provide information and support to people experiencing a range of issues including family problems, anxiety, depression, crisis and trauma. Visit our page on **Social Work** for more information.
- A genetic counsellor can help you understand how BMD is inherited and whether there are any implications for other members of your family. Your specialist will be able to advise whether a genetic counsellor is required. Visit our page on **Genetic Counselling** for more information.
- Your state or territory neuromuscular organisation can provide support, advice and information about living with DMD. Visit **state or territory neuromuscular organisations** for more information.
- Your GP can talk to you about a mental health plan and how you can use this to help you access the support and services you need. Visit our page on **Wellbeing** for more information.
- Other people and families living with BMD have also experienced the emotional rollercoaster that comes with a diagnosis of BMD and are able to understand exactly how you're feeling. You are not alone. Connect with other people and families living with BMD in The Loop Community on our **Forum**.

How to talk to your child and others about a new diagnosis

Adults can be diagnosed with the condition for the first time, but often patients are diagnosed when they are a child or an adolescent. Having a conversation with your child about their diagnosis is not easy. In fact it will probably be one of the hardest talks you will have. But, like all difficult conversations, it is important and necessary.

There are a lot of reasons why parents may not want to have this conversation, mostly centred on wanting to protect their child.

But avoiding the topic with your child is not helpful. Children are often aware of the differences between themselves and their siblings and/or peers and may hear their condition being discussed during medical appointments.

So it is crucial that you have a role in your child learning about their diagnosis. This will enable you to support your child as they process the information and to be on hand to answer any questions your child may have.

It will also allow you to:

- provide answers to questions in an age appropriate way
- correct any misinformation your child has heard or read
- show your child that you have faith in their ability to handle difficult conversations.

Ultimately, these conversations are an important step in providing your child with the tools to succeed, navigate their world and develop independence and self-advocacy skills.

There is no right time to have this conversation. Research suggests the earlier you talk to your child about their condition, the more natural the conversation will become. The important part is to pick a time and commit to it.

Some important do's and don'ts:

- Do be positive but also realistic
- Do reassure them that they have done nothing wrong and this is not their fault
- Do tell them they will do many wonderful things in their lives – they may just do them differently
- Do tell the truth. Answer all their questions.
- Do consider your language. Try to avoid negative or emotive words, such as 'suffering from a condition'.
- Do use daily living examples that they can relate to
- Don't avoid answering questions and don't shut the conversation down.

If you are having difficulty starting the conversation, try asking a couple of questions such as 'How are you feeling today?' or 'Do you know why you are tired?'

Remember, it is okay to be upset during the conversation but try to avoid breaking down as this will only cause your child greater distress. Children are resilient and they generally handle information of this nature far better than adults.

Understanding BMD and how it's diagnosed

About BMD

Becker Muscular Dystrophy (BMD) is a condition that causes progressive weakness of the skeletal muscles (the muscles that control movement). It also commonly affects heart muscle. BMD is genetic condition, meaning it is usually inherited through genes from parents. BMD is similar to another type of muscular dystrophy, Duchenne Muscular Dystrophy, but BMD has a later onset and milder symptoms.

What causes BMD?

BMD is caused by a change (also called a mutation) in a gene. The gene affected in BMD helps produce a protein (proteins are the building blocks of our muscles, organs and other tissues) called dystrophin. Dystrophin is usually found in our muscle cells. It works together with other proteins to strengthen muscle fibres and is important for forming healthy muscles. It also protects muscles from injury as they contract and relax. and helps repair damaged muscle cells. People with BMD produce faulty dystrophin which is too short and doesn't function normally. Without fully functional dystrophin, muscle cells become fragile and are more easily damaged, causing weakness of skeletal and cardiac (heart) muscles.

How is BMD diagnosed?

Diagnosing BMD can be complex due to the variation in the age of onset of the condition.

Generally, a GP would undertake a comprehensive physical assessment and consider a patient's history as well as their family history. The GP may request a blood test test to check the amount of an enzyme called creatine kinase (CK) in your or your child's blood. CK often leaks from damaged muscle cells into the blood. High levels of CK in the blood can suggest a muscle problem but does not confirm BMD.

If this test finds raised CK levels a referral to a neurologist (a doctor who specialises in brain and nerve conditions) to do a thorough assessment is necessary.

The neurologist may request genetic testing to look for changes in the dystrophin gene. This can be a 2 stage test with initial results taking 8 – 12 weeks and a further 16 weeks for more detailed testing. This test can be done through the public hospital

system at no cost but will involve a fee if done privately.

A Geneticist (a doctor who specialises in genes) may also be involved to discuss the implications of the test results for you and your family and to arrange testing for extended family members. Further information about genetic testing can be found on the **Australian Genomics Health Alliance website**. During this time your child or yourself may be asked to see a specialist physiotherapist to test movement. Visit our page on **Physiotherapy** for more information.

BMD is closely related to Duchenne muscular dystrophy (DMD) as they are caused by a mutation to the same gene (dystrophin gene). There can be some differences in the mutation in BMD compared to DMD, however it can sometimes be difficult to determine the severity based on the genetic mutation alone. Further assessments, including MRI of muscle and occasionally muscle biopsy may be required. These investigations need to be done in consultation with a neurologist.

The genetics of BMD

BMD is a genetic disease, meaning it is caused by an error in one of our genes – the dystrophin gene. In BMD, some of these mistakes are inherited (passed down from parents to their children) and there may often be a family member who has the condition. But gene changes can also happen randomly in the child (spontaneous mutations) with no family history.

BMD is inherited in an X-linked pattern because the dystrophin gene that can carry a BMD-causing mutation is on the X chromosome. Every boy inherits an X chromosome from his mother and a Y chromosome from his father, which is what makes him male. Girls get two X chromosomes, one from each parent. Each son born to a woman with a dystrophin mutation on one of her two X chromosomes has a 50 percent chance of inheriting the flawed gene and having BMD. Each of her daughters has a 50 percent chance of inheriting the mutation and being a *carrier*. Most carriers have no symptoms of BMD however some carriers will experience symptoms such as muscle pain or weakness and are at a higher than average risk of developing heart problems. Carriers should undergo a complete cardiac evaluation in late adolescence or early adulthood, or sooner if symptoms occur, and should be evaluated every 5 years.

For more detailed information about how BMD is inherited visit **Your Genes, Your Health**.

Long-term outlook

Living with BMD usually means life, and the future, looks different from what you had planned. However children and adults with BMD can lead fulfilling, rewarding lives just like everyone else. School, friendships, sports and recreation, arts, university, work and travel are all possible when you have BMD. Medications, equipment, occupational therapy and physiotherapy can help manage some symptoms and improve the quality of life for people with BMD.

For more information about living with BMD, overcoming some of the day-to-day challenges and where to get the right support, visit:

- **Living Life**
- **Counselling**
- **Social Work**
- **Psychology**

The effects of BMD

Learning about BMD and surrounding yourself with the right healthcare providers, services and support can help you feel more in control of what lies ahead. Although every person's journey with BMD will be unique, the information below will help you understand this journey and how to prepare and plan for changes in the future.

What are the main symptoms of BMD?

The symptoms of BMD can begin at any time from childhood into the early adulthood but muscle weakness usually becomes apparent between the ages of 5 and 15 years. The symptoms of BMD can be very different from person to person, including the age of onset, severity and how quickly the disease progresses. BMD usually affects males but, in rare cases, can also affect females.

BMD affects skeletal muscles, the muscles that cause movement. Muscle weakness often begins in the legs and pelvic (hip) area and people with BMD might notice difficulty walking, running, hopping and jumping. Falls may become frequent. Some people with BMD will have problems getting up from the floor and may "walk" their hands up their thighs (Gower's manoeuvre)

in order to stand up. The calf muscles in the lower legs may become enlarged (pseudohypertrophy). Muscle weakness slowly progresses to the muscles of the shoulders, neck and arms.

In some cases, the first sign of BMD may be a heart condition called dilated cardiomyopathy. This form of heart disease enlarges and weakens the heart muscle, preventing it from pumping blood efficiently. There are medications and treatments for dilated cardiomyopathy however it can progress rapidly and be life threatening.

Other symptoms of BMD can include:

- fatigue (tiredness)
- loss of balance and coordination
- breathing problems and difficulty coughing due to weakening respiratory muscles
- learning difficulties, such as issues with attention, verbal learning and memory and emotional interaction, affecting a small percentage of people with BMD.

Community Advice

Hear from a community member who has walked the path before you.

The video is available at **https://youtu.be/_6oJ9Mc0UO8.**

Life stuff

To find out more about living life with a neuromuscular condition and to access stories and peer-advice from the community, visit our **Living Life** section.

Where to find more information about living with BMD

Here is a collated list of credible and useful information about living with BMD.

Becker Muscular Dystrophy fact sheet
From Muscular Dystrophy New South Wales, 2017. This fact sheet includes considerations for future planning.

What is Becker Muscular Dystrophy? A Guide for Parents and Families
From Muscular Dystrophy Association, United States, May 2019. This guide includes what are the signs and symptoms of BMD, what should I know about BMD, how is BMD treated and a glossary of terms.

Learning to Live with Neuromuscular Disease: A message for parents
The Duchess of Kent Children's Hospital at Sandy Bay. August 2017

My Beckers Story
A blog post from Brad Miller living in Ontario, Canada detailing his journey with BMD from his diagnosis at 10 years of age, through his teen years and life as an adult living with BMD.

Muscular Dystrophy Association (US)
This website has excellent information about muscular dystrophy and neuromuscular conditions. You can find a full list of neuromuscular conditions, symptoms, causes and care options, as well as use the clinical trial finder tool.

Management of BMD

There are a number of therapies and support that can be used to manage and monitor the symptoms of BMD including:

- Regular physiotherapy and hydrotherapy for maintaining physical strength and function and stretching programs to maintain range of movement in joints. Physiotherapy may also assist with use of manual cough techniques and use of cough assist machines when you get sick. Visit our page on **Physiotherapy** for more information.
- Prescription of orthotics (splints or shoe inserts), particularly ankle foot orthoses, to be worn at night to prevent stiffness and pain.

- Occupational therapy (OT) to help with activities of everyday living and equipment (eg. scooter or power wheelchair) and support for accessibility to home, school and the community. Visit our page on **Occupational Therapy** for more information.
- Pain relieving medications may be used for muscle pain or cramps.
- Regular heart monitoring and medications to manage heart complications. Visit our page on **Cardiology** for more information.
- Educational and psychological interventions to assist with learning difficulties and maintaining mental health. Visit our page on **Psychology** for more information.
- Respiratory management for monitoring of breathing muscles including sleep studies and monitoring for need to use non-invasive ventilation. Visit our page on **Respiratory Care** for more information.
- Steroid medication, such as prednisolone, may be prescribed in some people with BMD. These medications might help preserve muscle strength and function, prevent scoliosis (spinal curvature), and maintain walking ability. However, steroids also cause unwanted side effects such as increased appetite, weight gain, loss of bone mass and cataracts. The benefits and risks of steroids should be discussed with your specialist.
- Orthopaedic surgery (operations to correct bones, joints and muscles), including spine straightening surgery, may help make sitting, sleeping, and breathing more comfortable.
- Speech pathology can help with swallowing problems. Visit our page on **Speech Pathology** for more information.

Other helpful support services

The video is available at **https://youtu.be/BDaCNsd9R_s.**

- Your state or territory neuromuscular organisation can provide information and advice on what support they can offer such as local support groups, camps, programs, services in the local area, access to cough assist machines, advocacy or assistance in times of crisis. They can also provide an ear to listen if you need someone to talk to or guide you to get the assistance you are needing. Visit our page on **state and territory neuromuscular organisations** for more information.
- Seeing a psychologist, counsellor or social worker can be incredibly helpful if you, your child or other members of the family are having a tough time or struggling with negative thoughts and feelings. Learn more about what services are available and how to access them here. Visit our pages on **Counselling**, **Social Work**, **Psychology** and **Wellbeing** for more information.
- Living with BMD may mean that you or your child may need some assistance for everyday activities. Find out more about how a disability support worker could help and how to engage this type of support. Visit our page on **Disability Support Workers** for more information.
- Other people and families living with BMD have also experienced the emotional rollercoaster that comes with a diagnosis of BMD and are able to understand exactly how you're feeling. You are not alone. Connect with other people and families living with BMD in The Loop Community on our **Forum**.

Medical Alert Card

Muscular Dystrophy Queensland has developed a **Medical Alert Card for BMD**. A Medical Alert Card can help you communicate your care needs in a medical emergency situation. If you would like a Medical Alert Card to keep in your wallet or purse, please call (07) 3243 9700 or complete the form on the Muscular Dystrophy Queensland's **website.**

How to have better conversations when communicating your needs

To learn how to have better conversations when communicating your needs, visit the following pages:

Employers

Educators

Living Life: Education

About BMD

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What is Becker Muscular Dystrophy? A Guide for Parents and Families
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Parent Resource

Learning to Live with Neuromuscular Disease: A message for parents
The Duchess of Kent Children's Hospital at Sandy Bay. August 2017

Community Stories

My Beckers Story
A blog post from Brad Miller living in Ontario, Canada detailing his journey with BMD from his diagnosis at 10 years of age, through his teen years and life as an adult living with BMD.

The Loop's Living Life Section
For more stories written by the neuromuscular community, visit The Loop's Living Life section.

International Organisation

Muscular Dystrophy Association (US)
This website has excellent information about muscular dystrophy and neuromuscular conditions. You can find a full list of neuromuscular conditions, symptoms, causes and care options, as well as use the clinical trial finder tool.