Myopathy

This is a fact sheet for parents whose child has had a muscle biopsy that confirms a myopathy. Adults with a myopathy may also find it helpful.

What is a myopathy?
We look at the muscle biopsy under the microscope for evidence of damage. Some changes suggest that the problem is not with the muscle itself, but with the nerve that controls that muscle and we call this neurogenic. Other changes suggest that the problem is with the muscle itself, and this appearance is called myopathic. When certain specific features are present we use the term dystrophic rather than myopathic.

Why do biopsies?
We do muscle biopsies to be able to see what is going on in the muscle and to see if we can discover what is causing the problems that your child is experiencing. This will then allow us to be aware of any possible, future problems. In an abnormal muscle biopsy there is more variation in fibre size and more irregularities in the muscle fibre than in normal muscle. However, contrary to what is found in dystrophic processes, there are no significant signs of damage or scarring. This may be an important distinction because the kinds of conditions that are associated a myopathic process in the process are different to the kinds that are associated with dystrophic changes, which can indicate more severe muscle damage.

What causes a myopathy?
There are many different underlying causes of a myopathic biopsy. If a muscle looks myopathic, there may be special features present, like ‘cores’ or ‘rods’ in the muscle which then point to an even clearer diagnosis than just a ‘myopathy’, but in many cases there are no specific features or clues that will point to a particular type of myopathy. In these situations it can take us a long time to work out the exact cause of the problem.

Is it possible that in the future they may be a more specific diagnosis?
When we do not come up with an exact diagnosis straight away, we continue to look at the biopsy using more specialised tests, as and when they become available, this process may take several years. We can also learn from follow-up appointments how things develop at the clinical level, and this can give us clues as to what is happening to your child. Sometimes, later on, we need to do a further biopsy as this can occasionally show up features, which were not present on the initial biopsy, but this will need to be discussed at the time.
What problems should I be aware of?
It is frustrating for parents who, when planning their child’s future, do not know the exact cause of their child’s myopathy; however, there are the things we have to look after.

It is important to realise that not all of the following problems may come up in an individual child, but because there is a risk of these kinds of issues we must be aware and be careful to watch out for: Muscle weakness. The weakness in the muscles in patients with myopathy is often fairly non-progressive, but can lead to problems with the range of movement of the joints or with the back. This means that we need to keep a close eye on mobility and joints and we do this in conjunction with the local physiotherapy team as well as through the muscle clinic.

People with myopathies can have chest problems because their cough is not strong. Where this may be a problem, we will be recommending the flu and pneomax immunisations. We also keep a close eye on your child’s chest and teach some specialised physiotherapy techniques to help him cough. If your child becomes ill with a chest infection, it is important this is treated promptly.

Feeding difficulties can also be an issue, and if someone is not eating or feeding very well then that can lead to undernutrition. You will need to be aware of this. Some people with a myopathy can also have problems with constipation, which can be managed in the usual kinds of ways.

To ensure that a child is getting the help he or she needs, support and information to schools and other professionals may be necessary. You may have other questions about the long term implications of this condition on your child’s health. It is very difficult to generalise about this, for children with a myopathy may be very different from one another. This is something that is best discussed with your local doctor. As time goes by there may be other issues which come up and these will be all be dealt with, in partnership with the local services, when you come to the muscle clinic.

We’re here for you at the point of diagnosis and at every stage thereafter, and can:

▸ Give you accurate and up-to-date information about your or your child’s muscle-wasting condition, and let you know of progress in research
▸ Give you tips and advice about day-to-day life, written by people who know exactly what it’s like to live with a muscle-wasting condition
▸ Put you in touch with other families living with the same muscle-wasting condition, who can tell you about their experiences
▸ Tell you about – and help you get – the services, equipment and support you’re entitled to.
If you would like your GP or other health professional to have more information about myopathy, we have some relevant materials. We’ve developed an online training module for GPs, as well as one for physiotherapists working with adults with muscle-wasting conditions. Contact our helpline or email us to find out more.

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Here for you
The friendly staff in the care and support team at the Muscular Dystrophy UK’s London office are available on 0800 652 6352 or info@musculardystrophyuk.org from 8.30am to 6pm Monday to Friday to offer free information and emotional support.

If they can’t help you, they are more than happy to signpost you to specialist services close to you, or to other people who can help.

www.musculardystrophyuk.org