Juvenile dermatomyositis (JDM)

If you have any questions about juvenile dermatomyositis, do get in touch with our research team on research@musculardystrophyuk.org. If you'd like to speak to others with the same condition as yours, you can visit our active and friendly online forum. (You'll find contact details at the end of the factsheet.)

What is JDM?

Dermatomyositis is a condition that tends to affect the muscles and skin but can affect other parts of the body. It is an '**autoimmune**' condition, which means that the immune system, which usually protects the body against infection/germs, becomes overactive and starts attacking the body rather than protecting it.

If dermatomyositis starts before the age of 16 years, we call it '**juvenile'** (childhood) **dermatomyositis.** JDM is very rare; about four children in every million are diagnosed with it each year. Girls are twice as likely as boys to get it. JDM usually starts between the ages of four and 10, but can affect children of any age.

What are the causes of JDM?

As with most autoimmune conditions, the cause of JDM is unknown. Many doctors and researchers across the world are studying JDM in detail to find what causes it.

Some genetic (hereditary) changes are more common in children with JDM than children without it, but these do not explain the whole story. It is thought that autoimmune conditions start after contact with a 'trigger', which makes the body's immune system overreact. Research has not yet identified one specific trigger but it is thought that triggers could include viruses or an environmental factor (such as sunlight).

JDM occurs by chance. Some families may be prone to autoimmune conditions and therefore children with JDM may have relatives with other autoimmune conditions, such as diabetes, thyroid disease, lupus or arthritis. Since JDM is so rare, it is unlikely that two members of one family will have it. Likewise, it is unlikely that a person with JDM will go on to have children with JDM. JDM is also not contagious so it cannot be caught or passed on from one person to the next.

How is JDM diagnosed?

A specialist (such as a paediatric rheumatologist, neurologist or dermatologist) will make the diagnosis of JDM. The condition is so rare that most GPs will not have come across the condition.

What are the signs and symptoms of JDM?

Weak, painful muscles: the muscles that tend to be affected the most are those near the trunk (central part of the body), including the upper arms, thighs, neck and the trunk itself. However, JDM can affect any muscle in the body. Children may experience swallowing

difficulties or may develop changes in their voice (a 'nasal' voice). Children may have difficulty getting out of bed, climbing up stairs and getting up from the floor or a chair.

Skin rashes: the typical JDM rash usually occurs on the face, knuckles, elbows, knees and ankles. Rashes can become worse in sunlight (they are photosensitive). JDM rashes may not appear at the same time as the muscle weakness; they can appear before or even after. Commonly, a red or purplish rash appears over the eyelids, called a 'heliotrope rash'. On the face, the rash appears as a reddened area on both cheeks and can cross the nose – this is called a 'malar rash'. Across the knuckles, elbows and knees, the rash looks like red/violet, dry skin patches. These are called 'Gottron's patches'. The tiny blood vessels at the base of the fingernails may turn a pinkish colour, or may become more obvious. These are called 'nail-fold capillary changes'.

Fatigue, tiredness, irritability: children with JDM may tire easily. Exercise, such as walking or running, may become increasingly difficult. Concentration and memory may also be affected. Irritability is common in young children with this condition.

Other symptoms: some children may have:

- swelling in the face or body, particularly around the eyes owing to fluid building up in the tissues (this is called 'oedema' and is more common early on in the illness)
- calcinosis: small hard calcium lumps under the skin or in the muscle. These lumps can break through the skin and leak a thick milky white fluid. If this happens, there is a chance of the lumps becoming infected. Calcinosis is more common when the child has had the condition for a long time.
- wasting away of the fatty tissue under the skin (this is called lipodystrophy)
- joint pain ('arthralgia') and/or swelling or stiffness in the joints ('arthritis')
- fever (temperatures)
- mouth ulcers
- headaches
- hair thinning or hair loss ('alopecia')
- change of colour of the hands in the cold ('Raynaud's phenomenon')
- chest pain
- abdominal (tummy) pain and/or bowel problems (such as diarrhoea or blood in the stool)
- involvement of organs of the body (such as the heart, lungs, brain).

It is unlikely that children will experience all of the above – symptoms can vary from child to child. JDM can range from being mild, where children may have few symptoms and it barely affects them, to much more severe. JDM can start suddenly, but more often, it develops gradually, with children becoming unwell over time. Some children may have muscle inflammation (pain/weakness) without a rash or skin problems.

It is not unusual for children to have features of other autoimmune conditions, such as scleroderma (with tightness/thickening of the skin) or lupus (systemic lupus erythematosus) as part of their JDM. This is called an 'overlap' syndrome.

Blood tests

Taking blood tests over a period of time helps doctors to work out how the condition is progressing and to monitor possible side-effects of medicines. Although you may need to have many blood tests done, these can usually be done from one sample taken at each clinic visit. Your child can be offered a gel to numb the skin to prevent the needle causing pain. Common blood tests can include:

- **muscle enzymes (such as CK, LDH):** when muscles are inflamed, they can become 'leaky' so that muscle enzymes (substances that start chemical reactions) leak into the blood. Blood tests show that the muscle enzymes are high, particularly when the condition is newly diagnosed. Later on, however, it is possible for JDM to be active but the muscle enzymes to be normal.

- **liver function tests (AST, ALT):** although these tests are associated with problems in the liver, AST and ALT can also be high owing to muscle inflammation (even if the liver function is normal)

- **full blood count (FBC):** this counts the number of cells in a sample of blood, including:
    - white blood cells (immune cells that fight infection and have a role in inflammation)
    - platelets (a component in the blood that is important for clotting)
    - the level of haemoglobin (Hb – the protein that carries oxygen around the body) in the blood. (Hb can be low in JDM because of anaemia, whereas white cells and platelets can be high owing to inflammation.)

- **Anti-nuclear antibody test (ANA)**: antibodies are proteins produced by blood cells. They normally circulate in the blood to defend against foreign invaders, such as bacteria, viruses, and toxins. Sometimes the body can produce antibodies against its own cells. There are blood tests available that look for particular antibodies associated with JDM – these include ANA and antibodies that are more specific. AANA can be detected in JDM, but is not specific and may be seen in other autoimmune conditions. Likewise, somebody may not have these antibodies and still have JDM.

- **Myositis Specific Antibodies (MSA) and Myositis Associated Antibodies (MAA):** these tests, which until recently, were only available within research studies, are now more widely available in clinical practice but should be done in a laboratory with expertise in testing them. About 70 percent of children with JDM will have a positive MSA or MAA. When present, they can help identify certain clinical patterns (for example risk of worse skin disease, calcinosis, and arthritis or lung involvement).

To help with the diagnosis of JDM, you may have the following tests done:

- **MRI scan of muscles:** an MRI scan takes place in a large tube with powerful magnets. Your child will need to lie still in this tube for about 20-30 minutes so the

machine can take images of the thigh muscles. The scan does not hurt but the machine is quite noisy.

- **muscle biopsy:** it may be necessary to take a very small bit of muscle from the top of the leg to look at under a microscope. This usually takes place under a general anaesthetic (putting your child to sleep).

- **skin biopsy:** doctors may take a small sample of skin to look at under a microscope. They will do this at the same time as a muscle biopsy, under a general anaesthetic. Otherwise, they will do it at a different time using a local anaesthetic to numb the area of skin.

- **EMG:** this looks at the electrical activity of muscles by inserting small needles (called electrodes) into them. In recent years, it has become more common for doctors to use an MRI scan rather than an EMG to help diagnose JDM. However, an EMG can help in some cases to find out if the weakness is in the muscles (myopathy), as opposed to in the nerves (neuropathy) or in the junction between the nerves and the muscles (neuromuscular junction).

- **X-rays:** your child may have X-rays taken of the inside of their chest or joints. They can also have X-rays to look for calcinosis in the arms or legs.

- **CT scan:** some children will need a CT scan to take pictures of the inside of their chest. This involves lying still in a tube (a bit like an MRI) for a short period.

- **lung function tests:** these are to look at how well the lungs work. Your child will need to blow into a special machine.

- **swallowing tests:** some children will need to see a Speech and Language Therapist who will assess the way they swallow. In some cases, an X-ray test is used (called 'video fluoroscopy'). This involves swallowing some liquid while they have X-rays taken.

- **ECG and echocardiogram (ECHO):** these tests measure the function of the heart. An ECG monitors how the heart beats, by sticking some leads to the chest using sticky plasters. An ECHO involves taking an ultrasound of the heart.

- **abdominal ultrasound:** this scan looks at the tummy, and the gel can feel a bit cold on the tummy.

The doctor will look at the results of all of the tests to see if your child has JDM. If your child is diagnosed with JDM, they will need to start treatment. If they do not respond to treatment, the treatment may need to be changed, or your child may need further investigations. It is usually at specialist centres where children receive their diagnosis, as the team will be experienced in diagnosing and treating the condition.

Is there a treatment or cure?

JDM can be treated effectively and kept under control with medication. There are specialist

centres across the UK where a team of people will be there to look after your child. This team is likely to include doctors that specialise in JDM, specialist nurses and specialist physiotherapists. Your child may also see other professionals as part of a team including occupational therapists, physiotherapists, and psychologists.

There are medicines to help your child get better, such as treatments that aim to reduce inflammation in the body and to lessen the chances of JDM getting worse. When JDM is severe or affects your child's organs, your child will need treatment quickly. Even if the condition is very mild, medicines can prevent any problems in the future. Treatment may change from time to time, depending on how JDM is affecting your child. Many children will get completely better over time, but often children need to take medicine for several years.

It is very important that your child take the prescribed medicines regularly. Although you may not notice a difference if your child misses one dose, the medicines will not work as well if they miss several doses. Your child's specialist doctor or nurse will discuss the medicines in detail with you and can give you written information to refer to.

The medicines used to treat JDM – known as 'immunosuppressants' – decrease the activity of the immune system. There are different strengths of immunosuppressants – each drug suppresses the immune system to a greater or lesser degree. If your child is on a stronger immunosuppressant, they may be more at risk of infection. Seek medical advice immediately, if your child becomes unwell. Some vaccinations (live vaccines) cannot be given while on these medicines, but other vaccines are encouraged. Your child's specialist will be able to advise.

Children with JDM are also often treated with steroids, such as prednisolone. These work quickly, but your child may need to take them for several months or longer. It is important to wait for your doctor's guidance before reducing the steroid dose. Like all medicines, steroids have side-effects. Your child's doctor/specialist nurse will explain what these can be.

Your child should not stop taking Prednisolone (or any other steroid treatments) suddenly; this may make your child extremely sick. Treatment of JDM with steroids is nearly always in association with the use of other immunosuppressant drugs, called 'disease-modifying drugs'. The most common of these used is methotrexate, but there are many others and your specialist doctor or nurse will give you information on these. Other drugs used to treat JDM include Intravenous Immunoglobulin (IVIG), Mycophenolate Mofetil (MMF), cyclosporine and azathioprine. For severe or resistant disease, doctors may prescribe the following drugs for your child: rituximab, infliximab, adalimumab, or cyclophosphamide.

You can find out the latest news on JDM research by visiting our website at http://www.musculardystrophyuk.org/news/news/research

To find out more about the research that Muscular Dystrophy UK is funding please visit: http://www.musculardystrophyuk.org/progress-in-research/research-projects/current-grants/

If you have any questions, please email our research team at research@musculardystrophyuk.org

Other ways to manage living with JDM

As well as encouraging your child to take their medication, here are a few things you can do to manage their JDM:

- sun protection: sunlight can make a JDM rash worse or can trigger a flare-up (worsening) of the condition. Encourage your child to use sunscreen with a sun protection factor (SPF) of 50 or more. Apply this 30 minutes before going out (even on cloudy days) and do not forget to apply to the ears. Protect exposed areas with a hat and long-sleeved shirts. Sometimes artificial lighting, such as fluorescent, halogen and CFL lights may affect the rash, so avoid those where possible.

- tiredness: this is very common in JDM. Your child may have a limited amount of energy and so it is important that they pace themselves. A regular sleeping pattern and routine to your child's day will help.

For more information on exercise, physiotherapy, diet, and vaccinations refer to factsheets on the Muscular Dystrophy UK website.

What is the probable course of JDM?

The condition can be managed well with treatment, enabling your child to do all the activities they want to, including sport. Your child won't always need to take medicine. When the condition is gone, your child will be in 'remission'.

Some children will have just one episode of JDM, which may last for two to three years, and then it goes away (enters remission). Some children may have a more prolonged condition that can last many years ('chronic course').

Once JDM has been diagnosed, it is important for it to be treated swiftly and vigorously by specialists. There is a small chance that the JDM will come back even after long periods of remission, but it can be treated if this happens. If you are worried about the possibility of a flare-up, speak to your child's specialist.

With treatment, children with JDM are likely to gain normal muscle strength and stamina over time. In some cases, children may have complications because of damage in their muscles/skin or from calcinosis – this is less likely if they receive treatment early on. Children with a prolonged chronic course can be more at risk of complications.

The condition itself, or the medicines taken for it, can lead to a delay in growth and puberty. Some children will need to see a specialist in growth (an endocrinologist).

Your child should be able to have healthy children in the future, despite having had JDM. Some medicines, such as cyclophosphamide, can affect fertility, but not usually in the doses given to treat JDM. There are some medicines, such as methotrexate, that treat JDM and are not for use during pregnancy, as they can cause harm to an unborn child. It is important to use contraception while taking these medicines. Speak to your clinician if you'd like to discuss this in more detail.

Dermatomyositis, JDM and JPM

In adults with dermatomyositis, there can be an association with malignancy (cancerous cells). This does NOT tend to be the case in children with JDM. Children are less likely than adults are to have problems with their heart or lungs because of JDM. Adults may have positive blood tests (myositis-specific antibodies) that are not always seen in children (children may have different antibodies). These differences suggest that the underlying conditions are different. It is therefore important for children to see professionals that specialise in JDM.

In some cases, the rash in JDM can occur later but a small number of children never have skin problems; this condition is juvenile **polymyositis** (JPM).

Most of the information in this factsheet is relevant for both JDM and JPM. Patients with JPM tend to have more severe symptoms at onset, including more frequent falling episodes and higher levels of muscle enzyme (CPK) than patients with JDM. Scientists have recently recognised a new sub-type of autoimmune myopathy without a rash, called 'necrotising autoimmune myopathy'. Blood tests (myositis-specific antibodies) and muscle biopsy changes help in its diagnosis.

Your child's hospital doctor will carry out tests to make sure there are no other reasons for the muscle weakness and that your child does have JDM or JPM.

Other related publications

- *Inheritance and muscular dystrophies*
- *Diagnostic tests*
- *A guide for parents: children with muscle-wasting conditions*
- *Education guidelines*
- *Adaptations manual*
- *Pregnancy and fertility*
- *Prenatal testing and diagnosis*
- *Carrier testing and reproduction: your options*
- *Exercise advice for adults with muscle-wasting conditions*
- Muscular Dystrophy UK's TalkMD forum: www.musculardystrophyuk.org/talkmd/
- Joseph Patrick Trust (to apply for a grant for specialist equipment): www.musculardystrophyuk.org/jpt
- A list of specialist paediatric rheumatology centres looking after children with JDM can be found on the Juvenile Dermatomyositis Research Group website: www.juveniledermatomyositis.org.uk/

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 JDM, 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.

If you have feedback about this factsheet or would like to request any references used to produce it, please email info@musculardystrophyuk.org.

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