Frequently Asked Questions

Juvenile Dermatomyositis (JDM) - What does it mean for my child?

GENERAL

What is Juvenile Dermatomyositis?

- Dermatomyositis is an autoimmune disease. Autoimmune means that cells that normally protect the body from infection react differently and cause inflammation that can manifest with pain, redness and swelling.
- In dermatomyositis, the small blood vessels in muscle (**myositis**) and skin (**dermatitis**) are affected. This inflammation leads to the problems that your child may have experienced such as muscle weakness, muscle pain and skin rashes on the face, eyelids, knuckles, knees and elbows.
- Dermatomyositis is seen in children and in adults. If the symptoms appear before the age of 16 years, we call the disease '**Juvenile' Dermatomyositis (JDM).**
- Some children / young people may have muscle inflammation without a rash or skin problems. Rashes can occur later but some children / young people never have skin problems and their disease is called **Juvenile Polymyositis (JPM)**. Both Juvenile Dermatomyositis (JDM) and Juvenile Polymyositis (JPM) belong to a group of rare conditions known as **Idiopathic Inflammatory Myositis (IMM)**. "Idiopathic" means that we do not know exactly the causes of these diseases.
- The hospital doctor will carry out tests to confirm which disease your child/young person has.

How common is JDM?

- JDM is a very rare illness in children. It affects approximately 3 to 4 children in every million each year.
- Girls are twice as likely as boys to get it.
- The illness usually starts between the ages of four and ten, but children of any age can be affected.

What causes JDM?

- The cause of JDM is unknown. Doctors and researchers across the world are looking at JDM and are trying to find what causes it.
- There is likely to be a genetic (hereditary) predisposition to all autoimmune diseases. We know some of the genes (messages that are passed on from parents to children) are more common in children with JDM than children without the disease but these do not explain the whole story.
- We think autoimmune diseases start due to contact with a 'trigger', which makes the body's immune system overreact. Research has not yet identified one specific

trigger but it is thought they could include infections (viruses, bacteria) or an environmental factor (such as sunlight).

Why my child? Is it my fault?

JDM occurs by chance and there is nothing that you or your child have done (or not done) to cause it.

Is JDM an inherited disease?

- JDM is not an inherited disease in that it is not passed down directly from a mother or father to their child. Some genes (which are "messages" passed from parents to children) make a person more likely to develop an auto-immune disease but this does not mean that every individual with these genes will develop an autoimmune disease. Many factors can contribute to developing JDM.
- However, children with JDM may have relatives that have auto-immune diseases such as diabetes, thyroid disease, lupus or arthritis. It is very unlikely that two members of one family will have JDM and it is also very unlikely that a person with JDM will go on to have children with JDM.

Is it contagious?

JDM is not contagious so it cannot be caught or passed on from one person to another.

Can JDM be prevented?

The cause of JDM is not yet known so we do not know how or if JDM can be prevented. Research into JDM will possibly help to find the answers to the prevention and treatment of the disease.

Is dermatomyositis different in children than in adults?

- Yes, the disease in adults is often more severe than that in children.
- In adult dermatomyositis, there can be an association with malignancy (presence of cancerous cells), which has not been reported in the Juvenile form of the disease.
- Adults sometimes have positive blood tests (myositis specific antibodies) that have different associations from those found in children.
- Both in children and adults, the illness can start suddenly, however many children usually become unwell gradually. Differently than adults, children are unlikely to have problems with their heart or lungs due to JDM while calcinosis is more frequently seen in children.
- Polymyositis is more frequent in adults and rare in children.

- It is not unusual for children with JDM to have features of other auto-immune diseases like arthritis, lupus, and scleroderma as part of their JDM. This condition is called an 'overlap' syndrome and it is less common in adults.
- Because of these differences between adult and childhood dermatomyositis, it is important for children to be seen by professionals who specialise in looking after children with JDM.
- Myositis Specific Antibodies are found in children and adults but have different association.

SIGNS AND SYMPTOMS

What are the signs and symptoms of JDM?

- **Muscle weakness and pain:** Muscles near to the central part of the body, including the upper arms, thighs, neck and the trunk itself are mostly involved in JDM. However, JDM can affect any muscle in the body. Children may experience swallowing difficulties or may develop changes in their voice (that becomes more 'nasal'). Children may have difficulty getting up from bed, climbing up stairs and getting up from the floor or a chair.
- **Skin rashes:** The typical rash that we see in children with JDM usually occurs on the face, knuckles, elbows, knees and ankles. Rashes can become worse in sunlight. This means that they are photosensitive. JDM rashes may not appear at the same time as the muscle weakness, as they can appear before or even after. The following is a description of common rashes or other skin changes associated with JDM:
    - On the eyelids the rash appears as a red, purplish colour. This is 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 become more obvious. These are called '**nail-fold capillary changes.**'
    - Children may get swelling to the face or body, particularly around the eyes, due to watery fluid building up in the tissues. This is called **oedema** and when it occurs aroiund the eyes, it is called 'peri-orbital oedema'.
    - Some children can develop small hard lumps under the skin or in the muscle that are due to calcium being deposited. This is called '**calcinosis**.' These lumps can break through the skin and leak a thick milky white fluid (calcium) and may eventually become infected. Calcinosis can be a sign of active disease or ongoing inflammation. These lumps are more common when the disease has been there for a long time.
    - Sometimes fatty tissue can disappear in JDM. This is called **lipodystrophy.**

- **Fatigue, tiredness:** Children with JDM may become tired easily. Exercise or even walking may become increasingly more difficult. They may need to take frequent rests and it may become harder for them to keep up with their friends. Concentration and memory may also be affected.
- **Other possible symptoms**:
    - Irritability (commonly seen in children with active disease, particularly in younger ones).
    - Joint pain ('arthralgia') and / or swelling or stiffness in the joints ('arthritis').
    - Fever (high temperature).
    - Mouth ulcers.
    - Headaches.
    - Hair thinning or hair loss (alopecia).
    - Change of colour of the hands in the cold (Raynaud's phenomenon).
    - Breathing problems (such as chest pain, cough, shortness of breath).
    - Abdominal (tummy) pain and / or bowel problems (such as diarrhoea or blood in the stool).
    - Voice changes, difficulties with feeding or swallowing.

It is unlikely that your child will experience all of the above symptoms.

Is JDM the same in all children?

- JDM can be a very variable illness. It can range from being mild, where children may have a few symptoms, to a more severe disease, which can affect different organs of the body (such as the heart, lungs, brain) as well as the muscles and skin.
- When JDM affects children's organs such as heart, lungs, abdomen (gut) or brain, treatment needs to be given quickly.
- If a child has very mild disease, it is still usual to treat them with medicines to prevent any further complications and to switch off the disease activity.
- Sometimes the child does not have any muscle weakness at all and shows mainly skin changes (we call this **amyopathic dermatomyositis**). Others will just have muscle disease without any skin disease (**polymyositis**). Sometimes the rash can appear later than muscle weakness in JDM.

How can I tell how severe my child's disease is?

- The doctor looking after your child will be able to tell you how severe your child's illness.
- JDM is a rare disease, therefore your GP is unlikely to have had experience of this illness and so it is best for you to talk to your child's specialist.

- Most children with dermatomyositis will be under the care of a paediatric rheumatologist, neurologist or dermatologist.

Can children have JDM without having a rash?

Yes. Some children may just get muscle involvement without any skin rashes. We call this Juvenile Polymyositis.

INVESTIGATION/ DIAGNOSIS

What happens when my child is diagnosed with JDM?

A team of health professionals will be there to look after your child and help them to recover and manage their disease. This Specialist team will include doctors, nurses, physiotherapists, occupational therapists and more. They will look after your child along with your local children's healthcare team.

What laboratory tests are done in JDM?

- Blood tests are the first diagnostic step when JDM is suspected. They are used to monitor the illness and also the medicines that your child will be taking as well. Once your child is established on treatment, blood tests will become less frequent. Blood tests that are commonly taken in JDM are as follows:
    - **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 can show that the muscle enzymes are high, particularly early in the illness.
    - **Liver function tests (AST, ALT).**

      Although these tests are also associated with liver problems, they can also be high due to muscle inflammation.
    - **Full Blood Count (FBC).**

      This counts the number of cells in a sample of blood, including haemoglobin (that carries oxygen around the body), white cells (the blood cells that fight infection and are important in inflammation) and platelets (blood particles important in clotting). Haemoglobin (Hb) can be low in JDM (so called anaemia) whereas white cells and platelets can be high due to inflammation.
    - **Antibody tests**.

      Antibodies are proteins produced by blood cells which normally circulate in the blood stream to protect the body from bacteria and viruses. Sometimes, the body can produce antibodies against its own cells and this can happen in JDM. Therefore, we can do blood tests to look for particular antibodies associated with JDM (myositis specific antibodies), which are only available at

the moment as part of the research studies). Although other antibodies, like antinuclear antibodies (ANA) and myositis-associated antibodies (MAA) can be detected in the blood in JDM, they are not JDM specific and they may be seen in other auto-immune disease.

What other tests will be performed?

To help with the diagnosis of JDM, the following tests may also be done:

- **MRI scan of muscles:** An MRI scan consists of a large tube with powerful magnets. Your child will need to lie still in this tube for approximately 20-30 minutes for pictures to be taken of the thigh muscles. The scan does not hurt but it is quite noisy.
- **X rays:** These may be taken of the chest or joints. They may also be taken to look for calcinosis in the arms or legs
- **ECG and ECHO cardiogram:** These tests look at the function of the heart. Neither test hurts. An ECG involves sticking some leads to the chest using sticky plasters and monitoring how the heart beats. An echocardiogram involves an ultrasound of the heart. The scan does not hurt but the jelly can feel a bit cold.
- **Abdominal Ultrasound:** This is a scan of the tummy. The scan does not hurt, but the jelly may feel a bit cold.
- **Lung function tests:** These tests look at how well the lungs work. This is done by blowing into a special machine. It can be done only in older children.
- **CT scan**: Some children will need a CT scan of their chest. This involves lying still in a tube (a bit like an MRI) for a short period of time. It does not hurt.
- Some children will need a **Speech and Language Therapist** to assess the way that they swallow. In some cases, an X-ray test is used to assess swallowing (**video fluoroscopy**). This involves swallowing some liquid whilst x rays are taken.
- **Muscle biopsy:** a very small bit of muscle from the top of the leg is taken to look at under a microscope to help with disease diagnosis and management. This is usually done asleep under a general anaesthetic.
- **Skin biopsy:** A small sample of skin may be taken to look at under a microscope. This can usually be done at the same time as a muscle biopsy.
- **EMG:** This looks at the function of muscles. It is done by inserting small needles called electrodes into the muscles to measure its electricity. This investigation can be useful to distinguish JDM from some congenital muscle diseases, but it is not always needed in straightforward cases.

How often are tests done? What happens in clinics?

- Blood tests are usually carried out when attending clinic. Your child may need blood tests more frequently at the start of their illness but they are usually reduced to 8 weekly when treatment is established.

- When attending clinic your child will be seen by their doctor. They may also see a physiotherapist and a nurse. They will be asked questions about how they have been feeling since their last clinic appointment. The doctor will then examine them and will want to listen to their heart and lungs, feel their tummy and examine their joints. The doctor / physiotherapist / nurse will also want to see how strong they are and they may test muscle strength in the arms, legs, neck and tummy.
- If your child has a flare (worsening) of their disease, they may need to undergo more investigations such as an MRI, X-rays or investigations of their heart (ECG / ECHO), or lungs (lung function tests or a CT scan).

How can I be sure that it is definitely JDM?

The doctor will look at the results of all tests to work out if your child has JDM. If your child does have JDM, they will be started on treatment. Your child may need more investigations if they do not respond as well as expected to treatment, or if the treatment needs to be changed according to disease evolution.

TREATMENT

How will my child get better?

- There are medicines to help your child get better. The treatment is aimed at reducing inflammation in the body by switching off active disease but also to keep the disease into remission. The treatment that is given may change from time to time. This will depend on how JDM is affecting your child.
- It is very important that the prescribed medicines are taken regularly. Medicines for JDM work in the background over time and so although your child may not notice differences if one dose is missed, the medicines will not work as well if several doses are missed.
- Your child's doctor or nurse will explain which medicines can help your child and provide written information for you to read.
- Your child may require several medications to switch off or control the disease activity.
- Medicines used to treat JDM decrease the activity of the immune system and are called 'immunosuppressants'. Your child may be more at risk of infection and you should seek medical advice if your child becomes unwell. Live vaccinations cannot be given whilst on these medicines.
- Here are some of the medicines that may be used. ***NB: This is not a complete description of the medicines action and side effects and your child should be given information by your specialist doctor or nurse.***

Steroids:

Prednisolone: This medicine works quickly to decrease the inflammation (redness, pain, swelling) caused by JDM. It may be given as an infusion initially and then switched to oral medication. The steroids will be slowly weaned as other medication is used to control the disease. Side effects of prednisolone are usually related to the dose taken and how long it is taken for. It is important to wait until the doctor guides your child about reducing the steroid dose. Prednisolone should **NEVER** be stopped suddenly as this may make your child very unwell. The doctor will reduce your child's prednisolone gradually guided by your child's response to treatment.

Methylprednisolone: This drug is similar to prednisolone except it is given into a vein in your child's arm. It is often given in the early stages of JDM when disease is more active or may be given at times of increased disease activity called a flare.

Disease Modifying Anti-Rheumatic Drugs (DMARDS):

Treatment with steroids is usually associated with use of other drugs that treat JDM by suppressing (decreasing) the immune system. These drugs are called 'Disease Modifying Drugs' and the most common drug used in addition to steroids is methotrexate.

Methotrexate: This drug reduces inflammation by acting directly on the immune system. It takes 6 to 8 weeks to start working and is usually given over a long period time to maintain remission (keeping the disease quiet). Methotrexate can affect the white blood cells which fight infection, and also irritate the liver, so regular blood tests are required. Methotrexate has been used for many years and is usually the first line treatment given for most patients with JDM in addition to steroids.

Other medicines used to reduce the overactive immune system are:

Ciclosporin

Azathioprine

Mycophenolate Mofetil (MMF)

These medicines require blood tests to assess response to treatment and monitor for potential side effects on the blood cell count.

Hydroxychloroquine: This medication is used particularly for skin disease or joint aches and pains. If your child is taking hydroxychloroquine, they should have an eye test with an optician once a year since it can (rarely) cause changes in colour vision or peripheral vision (seeing objects outside the direct line of vision). This usually recovers once the drug is stopped.

Immunoglobulins (IVIG): This contains human antibodies (proteins) concentrated from blood. It is given through a vein in your child's arm. IVIG works through the immune system to reduce inflammation. It is usually given in addition to other medicines.

Cyclophosphamide: This is usually only given in more severe cases of JDM and when there is organ involvement (e.g. lungs, brain) or severe skin or muscle changes. It is a stronger medication to decrease the immune system activity and it is given through a

vein in your child's arm in hospital. Your child may require 6 to 7 doses of medication given over several months.

Biologic agents:

Infliximab: This is given through a vein in your child's arm. Like most of the other medicines this works by reducing inflammation and regular blood tests are required. Infliximab is a type of medication called a biologic that targets the immune system in a more specific way. It tends to be given as an extra treatment in those children that do not respond well enough to initial medication with drugs such as methotrexate.

Rituximab: This is also an immunosuppressive drug used in severe disease. It is given through a vein in your child's arm. Two doses are usually given two weeks apart and then may need to be repeated at a later date.

Adjuvant treatment:

Calcium and Vitamin D: Supplementation with calcium and Vitamin D is suggested especially when your child is on steroids to prevent osteoporosis.

Are there things that I can do to help my child's JDM?

There are things that you can do to help your child in addition to encourage them to take their medication

Sun protection: Sun protection is important because sunlight can make JDM rash worse or can trigger a flare (worsening) of the disease.

- Encourage your child to use sunscreen with a sun protection factor (SPF) of 50 or more. Be sure to apply 30 minutes before going out even on cloudy days and do not forget to use on the ears. Find a preparation that best suits your child as some rub in better than others.
- Protect exposed areas with a hat and long sleeved shirts.
- Sometimes the rash may be affected by artificial lighting such as fluorescent, halogen and LED lights, so do check whether lighting in the home or school could be affecting your child. If so, then it could be filtered or the bulb strength reduced.

Medications: It is important that your child remembers to take their medication as prescribed by the doctor, even when they start to feel better.

Fatigue and pacing: Fatigue (feeling tired) is very common in JDM. Your child may have limited energy and so it is important that they pace themselves and gradually increase activity over time.

Exercise: Exercise is always important, but during a flare of JDM it may be very difficult to do exercise. Common symptoms of JDM are muscle weakness and joint stiffness, which may reduce your child's mobility and fitness. Shortening of affecting muscles can lead to restriction in movement.

Physiotherapy is important to improve muscle strength and increase activity.

The physiotherapist will teach your child exercises designed to

- build up muscle strength and stamina (sustaining energy or strength over a long period), and
- improve and maintain the range of movement of their joints.

You can help by encouraging your child to do their physiotherapy exercises. Once your child is feeling better, it is important to get back to doing regular exercise and pacing activities to gradually increase exercise tolerance.

Diet: No special diet will cure or prevent a flare of JDM. A well- balanced diet that includes a variety of foods is recommended. Steroids can make your child feel hungry so try to encourage healthy small snacks such as fruit, vegetables, nuts and seeds to reduce weight gain.

Sleep: Your child may feel more tired than usual due to JDM. This will improve over time with treatment. A regular sleeping pattern and daily routine will help.

Vaccinations: When your child is taking immune-suppressive medications, they will not be able to receive 'live 'vaccinations such as Measles Mumps and Rubella, Chicken Pox, Tuberculosis and some travel vaccinations. There are many non-live vaccinations that your child can still receive and it is recommended that a flu vaccination (flu jab) is given each year. Children should also have a pneumococcal vaccination (to prevent pneumonia) every 5 years. You can ask your GP or specialist doctor or nurse about any vaccinations that you are not sure about.

How long are children with JDM on treatment for?

- Each child / young person is different and the length of drug treatment will depend on the characteristics of the disease in the individual child and the response to treatment. For some children the disease may be short lived, whilst others may have the disease for many years.
- Doctors aim to control the disease so that your child can lead a normal life and do everything that they wish to do. Treatment would usually only be stopped after your child has not had any symptoms (complaints) of JDM for some time (usually at least 1 year with no complaints). JDM is a disease particularly sensitive to drug treatment reductions. This means that if drugs are reduced too fast, it can cause a flare of the disease. Your child should only reduce medication on the advice of your specialist team.

Are there any alternative/ complimentary therapies for JDM?

Complementary or alternative therapies are those that fall outside the conventional medicines. These therapies can be based on cultural or historical traditions rather than scientific evidence. Some complimentary therapies are safe and can be used in addition to your child's medication but others may interact with prescribed drugs and increase the risk of side effects. It is highly recommended that you seek the advice of your specialist before using complimentary or alternative medications.

PROGNOSIS (LONG TERM OUTLOOK)

What is the long term outlook for children with JDM?

- Some children will have just one episode of JDM, which may last for 2-3 years and then goes away (enters what is called 'remission'). Other children will have disease that comes back after a prolonged period of remission. Some children have a more prolonged disease than can last many years ('chronic course').
- It is important for the disease to be treated aggressively by specialists in JDM. When this is done, the outlook is overall favourable.

Will this ever go away?

When the disease is quiet, we call this remission. Remission will occur with proper treatment so that your child will be able to carry out all of the activities that they wish to do. There is a small chance that the disease will come back even after long periods of remission, but it can still be treated if this happens.

Will the rash ever go away?

Yes, the rash will go away. Often the rash takes longer to go away than the muscle weakness.

Will I know if my child is having a flare (worsening) of JDM? How?

Your child may feel weak, have a rash or feel generally unwell. Young children will often become very irritable when they are having a flare (worsening) of their JDM. If you are worried about the possibility of a flare, you should contact your child's specialist team.

How will JDM affect my child in school?

- When the disease is active, your child may become more tired than usual at school and may find it difficult to concentrate.
- Muscle weakness can make it difficult to walk around school/ college. During active disease, your child may need to have shorter days at school/college as well as miss PE, but in time, this will increase to full days and increased exercise and activity.

Can my child get extra help from school/ college if needed?

- Schools / colleges can help to make it easier for your child to join in normally. When not feeling well, it may be advisable to focus on core subjects, reduce the workload, give time to catch up or rest, and provide extra time to get to classes and complete homework.

- Your child's medical team can help by talking to the teachers at school / college with any areas of difficulty. With your permission your child's medical team can write to teachers and provide more information about JDM.
- Lots of children/ young people with JDM have gone on to university and / or have successful jobs.
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How will JDM affect my child and my family's life?

A chronic disease like JDM is a challenge for the whole family. It is very important that parents are coping well so they are able to support and encourage the child with their positive attitude. When needed, psychological support should be offered by the paediatric rheumatology team

Is there anything that children with JDM will not be able to do?

Initially your child may struggle to continue with normal everyday activities. It may take a while for your child to get 'back to normal'. With all of the medicines and therapies discussed, we would hope that over time your child would be able to lead a normal life carrying out all of the activities that they wish to.

Will my child's puberty or growth be affected by JDM or by the treatments that they will be on?

- Growth and puberty can both be affected by active inflammation (pain, redness, swelling). The best way of improving growth is to control the disease.
- Steroids can also affect growth, but doctors will ensure that your child is treated with the lowest dose possible. During chronic illness such as JDM, puberty often occurs a little later than usual. If this happens, it actually means that your child will have more time to grow! Some children or young people may see a specialist in growth (an endocrinologist) if delay is longer than expected.

Will my child be able to have a family in the future?

- Your child should be able to have a healthy child in the future, despite having had JDM. Some people cannot have children for other reasons. Some medicines (such as cyclophosphamide) can affect fertility. However, this only occurs when large doses are given. If cyclophosphamide is given before puberty, there is less risk to your child's fertility.
- There are some medicines used to treat JDM that should not be taken whilst pregnant as they may damage an unborn child. It is therefore important that the young person talks to their doctor if they wish to become pregnant. Young people who are sexually active should use contraceptives to avoid pregnancy or fathering a child whilst they are taking these medicines.

What is in the future for my child?

- In some cases, children may have problems due to damage in their muscles/ skin or calcinosis, but this is less likely if children are treated early in their disease. Children with a prolonged chronic course can be more at risk of complications.
- Your child should be able to lead a normal life, firstly taking medication and in time, without medicine. We would expect children to return to normal muscle strength and stamina over time and be able to carry out all activities, including sporting activities.

Can we meet other families that have children with JDM?

JDM is very rare. It only affects approximately 3-4 children in every million each year. However, your specialist will be treating other children with JDM. If you would like to meet them or talk to them, ask your specialist.

Information leaflet prepared 2012 by CNS Louise Hanna, Dr Eleanor Heaf & Dr Liza McCann @ Alder Hey Children's NHS Foundation Trust on behalf of the JDRG (ratified by Steering Committee). Updated March 2016.