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# Juvenile Dermatomyositis

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## 1. WHAT IS JUVENILE DERMATOMYOSITIS

### 1.1 What kind of disease is it?

Juvenile dermatomyositis (JDM) is a rare disease that affects muscles and skin. A disease is defined as "juvenile" when it starts before the age of 16.

Juvenile dermatomyositis belongs to a group of conditions that are thought to be autoimmune diseases. Usually the immune system helps us to fight infections. In autoimmune diseases, the immune system reacts in a different way and becomes overactive in normal tissue. This immune system reaction leads to inflammation, which causes tissues to swell and can lead to possible tissue damage.

In JDM, the small blood vessels in the skin (dermato-) and the muscles (myositis) are affected. This leads to problems such as muscle weakness or pain, especially in the muscles of the trunk and those around the hips, shoulders and neck. Most patients have typical skin rashes as well. These rashes can affect several areas of the body: face, eyelids, knuckles, knees and elbows. The skin rash does not always occur at the same time as the muscle weakness: it can develop before or after it. In rare cases, small blood vessels in other organs can also be involved.

Children, adolescents and adults can all develop dermatomyositis. There are some differences between adult and juvenile dermatomyositis. In ~30% of adults with dermatomyositis, there is a relation to cancer (=malignancy), whereas in JDM there is no association with cancer.

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## **1.2 How common is it?**

JDM is a rare disease in children. Approximately 4 in 1 million children will develop JDM each year. It is more common in girls than in boys. It most often starts between the ages of 4 and 10 years, but children of any age can develop JDM. Children from all around the world and from all ethnic backgrounds can develop JDM.

## **1.3 What are the causes of the disease and is it inherited? Why does my child have this disease and can it be prevented?**

The exact cause of dermatomyositis is not known. Internationally, there is a lot of research going on trying to find the cause of JDM.

JDM is currently regarded as an autoimmune disease and is probably caused by several factors. These could include a person's genetic predisposition in combination with exposure to environmental triggers such as UV-radiation or infections. Studies have shown that some germs (viruses and bacteria) can trigger the immune system to react abnormally. Some families with children affected by JDM suffer from other autoimmune diseases (diabetes or arthritis, for example). However, the risk of a second family member developing JDM is not increased.

Presently, there is nothing we can do to prevent JDM. Most importantly, there is nothing you could have done as a parent to prevent your child from getting JDM.

## **1.4 Is it infectious?**

JDM is not infectious, nor is it contagious.

## **1.5 What are the main symptoms?**

Each person with JDM will have different symptoms. Most children have:

### **Fatigue (tiredness)**

Children are often tired. This may lead to a limited capacity to do exercise and eventually to potential difficulties in day-to-day activities.

### **Muscle pains and weakness**

Muscles close to the trunk are often involved, as well as muscles in the

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abdomen, back and neck. In practical terms, a child might start to refuse walking longer distances and doing sports, small children may "become fussy", asking to be carried around more. As the JDM gets worse, climbing stairs and getting out of bed might become a problem. In some children, the inflamed muscles become tight and shorten (called contractures). This leads to difficulties in fully straightening the affected arm or leg: the elbows and knees tend to be in a fixed bent position. This can affect the movements of the arms or legs.

### **Joint pain and sometimes joint swelling and stiffness**

Both large and small joints can be inflamed in JDM. This inflammation can cause swollen joints as well as pain and difficulty in moving the joint. This inflammation responds well to treatment and it is uncommon for it to result in damage to the joints.

### **Skin rashes**

The rashes seen in JDM can affect the face with swelling around the eyes (periorbital oedema) and a purple-pink discolouration of the eyelids (heliotrope rash); there can also be redness over the cheeks (malar rash) as well as on other parts of the body (top of the knuckles, knees and elbows) where the skin can become thickened (Gottron's papules). Skin rashes can develop long before muscle pain or weakness. Children with JDM may develop many other rashes. Sometimes doctors can see swollen blood vessels (appearing as red dots) in the child's nail beds or on their eyelids. Some JDM rashes are sensitive to sunlight (photosensitive), whereas others can result in ulcers (sores).

### **Calcinosis**

Hard lumps under the skin containing calcium may develop during the course of the disease. This is called calcinosis. Sometimes it is already present at the onset of disease. Sores may develop on top of the lumps and a milky liquid made of calcium can drain out. Once they have developed they are difficult to treat.

### **Abdominal pain or tummy ache**

Some children have problems with their bowels. These can include tummy aches or constipation, and occasionally severe abdominal problems if the blood vessels to the gut become affected.

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## **Lung involvement**

Breathing problems may occur because of muscle weakness. Muscle weakness can also cause changes in a child's voice, as well as swallowing difficulties. Sometimes there is inflammation of the lungs, which can result in shortness of breath.

In the most severe forms, virtually all muscles attached to the skeleton (skeletal muscles) may be affected, which results in problems with breathing, swallowing and speaking. Therefore, voice changes, difficulties with feeding or swallowing, coughing and shortness of breath are important signs.

### **1.6 Is the disease the same in every child?**

The severity of the disease varies with each child. Some children may have just their skin affected with no muscle weakness (dermatomyositis sine myositis), or with very mild muscle weakness that may only be apparent on testing. Other children may have problems with many parts of their body affected: skin, muscles, joints, lungs and intestines.

## **2. DIAGNOSIS AND THERAPY**

### **2.1 Is it different in children compared to adults?**

In adults, dermatomyositis can be secondary to underlying cancers (malignancies). In juvenile JDM, there is no association with cancer. In adults, there is a condition where just the muscles are affected (polymyositis) but this is very rare in children. Adults sometimes have specific antibodies detected by testing. Many of these are not seen in children, but specific antibodies have become recognised in children within the last 5 years. Calcinosis is more frequently seen in children than in adults.

### **2.2 How is it diagnosed? What are the tests?**

Your child will need a physical examination, along with blood tests and other tests such as an MRI or muscle biopsy to diagnose JDM. Each child is different and your doctor will decide on the best tests for each child. JDM can present with a specific pattern of muscle weakness (involvement of muscles in the thighs and upper arms) and specific skin

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rashes: in these cases JDM is easier to diagnose. The physical examination will include checking muscle strength, skin rashes and the blood vessels in the nail beds.

Sometimes JDM can look like other autoimmune disease (such as arthritis, Systemic Lupus Erythematosus, or vasculitis) or like a congenital muscle disease. The tests will help work out which disease your child has.

### **Blood tests**

Blood tests are performed to look for inflammation, immune system function and problems secondary to the inflammation, such as leaky muscles. In most children with JDM, the muscles become "leaky". This means there are substances in the muscle cells that leak into the blood, where they can be measured. The most important of these are the proteins called muscle enzymes. Blood tests are commonly used to assess how active the disease is and to assess the response to treatment at follow-up (see below). There are five muscle enzymes that can be measured: CK, LDH, AST, ALT and aldolase. The level of at least one of them is elevated in most patients, though not always. Other laboratory tests can help in the diagnosis. These include antinuclear antibodies (ANA), myositis-specific antibodies (MSA) and myositis-associated antibodies (MAA). ANA and MAA may be positive in other autoimmune diseases.

### **MRI**

Muscle inflammation can be seen using magnetic resonance techniques (MRI).

### **Other muscle tests**

The findings in a muscle biopsy (the removal of small pieces of muscle) are important to confirm the diagnosis. In addition, a biopsy can be a research tool for better understanding the disease.

The functional changes in the muscle can be measured with special electrodes that can be inserted as needles into the muscles (electromyography, EMG). This investigation can be useful to distinguish JDM from some congenital muscle diseases, but it is not always needed in straightforward cases.

### **Other tests**

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Other tests can be performed to detect involvement of other organs. Electrocardiography (ECG) and heart ultrasound (ECHO) are useful for heart disease, while chest X-rays or CT scan together with pulmonary function tests may reveal lung involvement. X-ray of the swallowing process using a special opaque liquid (contrast medium) detects involvement of muscles in the throat and oesophagus. Ultrasound of the abdomen may be used for gut involvement.

### **2.3 What is the importance of the tests?**

Typical cases of JDM can be diagnosed from the pattern of the muscle weakness (involvement of muscles in thighs and upper arms) and the classic skin rashes. Tests are then used to confirm the diagnosis of JDM and to monitor treatment. Muscle disease in JDM can be assessed by standardized muscle testing scores (childhood myositis assessment scale, CMAS; Manual Muscle Testing 8, MMT8) and blood tests (looking for elevated muscle enzymes and inflammation).

### **2.4 Therapy**

JDM is a treatable disease. There is no cure but the aim of treatment is to control the disease (get the disease into remission). The treatment is tailored to the needs of the individual child. If the disease is not controlled, then damage may occur and can be irreversible: it can produce long-term problems, including disability, which persists even when the disease has gone.

In many children, physiotherapy is an important element of treatment; some children and their families also need psychological support to cope with the illness and its effect on their daily lives.

### **2.5 What are the treatments?**

All medications work by suppressing the immune system, to stop the inflammation and prevent damage.

#### **Corticosteroids**

These drugs are excellent for controlling inflammation quickly. Sometimes corticosteroids are given via a vein (through an intravenous or IV line) to get the medication into the body quickly. This can be

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lifesaving.

However, there are side effects if high doses are needed long-term. The side effects of corticosteroids include problems with growth, increased risk of infection, high blood pressure and osteoporosis (thinning of the bones). Corticosteroids cause few problems at a low dose; most problems are seen with higher doses. Corticosteroids suppress the body's own steroids (cortisol), and this can cause serious, even life-threatening problems, if the medication is suddenly stopped. That is why corticosteroids need to be reduced slowly. In combination with corticosteroids, other immune suppressive medication (such as methotrexate) may be initiated to help control the inflammation long-term. For more information, see drug therapy.

### **Methotrexate**

This drug takes 6 to 8 weeks to start working and is usually given over a long period of time. Its main side effect is feeling sick (nausea) around the time it is given. Occasionally mouth ulcers, mild thinning of the hair, a drop in white blood cells or a rise in liver enzymes may develop. The liver problems are mild but they can be made much worse by alcohol. Adding folic or folinic acid, a vitamin, diminishes the risk of side effects especially on liver function. There is a theoretical increased risk of infections, although in practice, problems have not been seen except with chickenpox. While on treatment, pregnancy must be avoided because of the effects of methotrexate on the foetus.

If the disease is not controlled by the combination of corticosteroids and methotrexate, several other therapies are possible, often in combination.

### **Other immunosuppressive drugs**

Cyclosporin, like methotrexate, is usually given over a long period of time. Its long-term side effects include raised blood pressure, increase in body hair, gum enlargement and kidney problems. Mycophenolate mofetyl is also used long-term. It is generally well tolerated. Its main side effects are abdominal pain, diarrhoea and an increased risk of infections. Cyclophosphamide may be indicated in severe cases or in disease resistant to treatment.

### **Intravenous Immunoglobulin (IVIG)**

This contains human antibodies concentrated from blood. It is given into a vein and works in some patients through effects on the immune

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system, causing less inflammation. The exact mechanism of how it works is unknown.

### **Physiotherapy and exercise**

Common physical symptoms of JDM are muscle weakness and joint stiffness, resulting in reduced mobility and fitness. Shortening of affected muscles can lead to restriction in movement. These problems can be helped through regular physiotherapy sessions. The physiotherapist will teach both children and parents a series of appropriate stretching, strengthening and fitness exercises. The aim of treatment is to build up muscle strength and stamina, and to improve and maintain the range of movement of the joints. It is extremely important that parents are involved in this process to help their child maintain the exercise program.

### **Adjuvant treatments**

Correct intake of calcium and vitamin D is recommended.

### **2.6 How long should treatment last?**

The length of treatment is different for each child. It will depend on how the JDM is affecting the child. Most children with JDM have treatment for at least 1-2 years, but some children will need treatment for many years. The aim of treatment is to control the disease. Treatment may be gradually reduced and stopped once the child has had inactive JDM for a period of time (usually many months). Inactive JDM is defined in a child who is well with no signs of active disease and normal blood tests. Assessment of inactive disease is a careful process in which all aspects need to be considered.

### **2.7 What about unconventional or complementary therapies?**

There are many complementary and alternative therapies available and this can be confusing for patients and their families. Most therapies are not proven to be effective. Think carefully about the risks and benefits of trying these therapies as there is little proven benefit and they can be costly in terms of money, time and burden to the child. If you want to explore complementary and alternative therapies, it is wise to discuss these options with your paediatric rheumatologist. Some

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therapies can interact with conventional medications. Most doctors will not be opposed to complementary therapies, provided you follow medical advice. It is very important not to stop taking your prescribed medications. When medications, such as corticosteroids, are needed to keep JDM under control, it can be very dangerous to stop taking them if the disease is still active. Please discuss concerns about medication with your child's doctor.

## **2.8 Check-ups**

Regular checks are important. At these visits, JDM disease activity and potential side effects of the treatment will be monitored. As JDM can affect many parts of the body, the doctor will need to examine the whole child carefully. Sometimes special measures of muscle strength are done. A blood test is often required to look for JDM disease activity and to monitor treatment.

## **2.9 Prognosis (this means long-term outcome for the child)**

JDM generally follows 3 paths:

JDM with a monocyclic course: just one episode of disease that goes into remission (i.e. no disease activity) within 2 years after onset, without relapses; JDM with a polycyclic course: there may be long periods of remission (no disease activity and the child is well) alternating with periods of JDM relapses, which often occur when treatment is reduced or stopped; Chronic active disease: this is characterized by ongoing active JDM despite treatment (chronic remittent disease course); this last group has a higher risk of complications. Compared to adults with dermatomyositis, children with JDM generally do better and do not develop cancers (malignancy). In children with JDM who have internal organs affected, such as lung, heart, nervous system or gut, the disease is much more serious. JDM can be life-threatening but this depends on how severe the disease is, including the severity of the muscle inflammation, which organs of the body are affected and whether there is calcinosis (calcium lumps under the skin). Long-term problems can be caused by tight muscles (contractures), loss of muscle bulk and calcinosis.

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## **3. EVERYDAY LIFE**

### **3.1 How might the disease affect my child and my family's daily life?**

Attention should be paid to the psychological impact of the disease on children and their families. A chronic disease like JDM is a difficult challenge for the whole family and of course, the more serious the disease, the harder it is to cope with. It will be difficult for a child to cope properly with the disease if their parents are having problems coping. A positive attitude from parents to support and encourage a child to be independent as much as possible, despite the disease, is extremely valuable. It helps children to overcome the difficulties related to the disease, to cope successfully with peers and to become independent and balanced. When needed, psychosocial support should be offered by the paediatric rheumatology team.

Allowing the child to have a normal adult life is one of the main goals of therapy and it can be reached in the majority of cases. The treatment of JDM has improved dramatically in the last ten years and it is conceivable that several new drugs will be available in the near future. The combined use of pharmacological treatment and rehabilitation is now able to prevent or limit muscle damage in most patients.

### **3.2 Can exercise and physical therapy help my child?**

The purpose of exercise and physical therapy is to help the child to participate as fully as possible in all the normal daily activities of life, and to fulfil their potential within society. Exercise and physical therapy can also be used to encourage active healthy living. To be able to achieve these goals, healthy muscles are needed. Exercise and therapy can be used to achieve better muscle flexibility, muscle strength, coordination and endurance (stamina). These aspects of musculoskeletal health allow children to successfully and safely engage in school activities, as well as activities outside school such as leisure time activities and sports. Treatment and home exercise programs can be helpful to reach normal fitness level.

### **3.3 Can my child play sports?**

Playing sports is an essential aspect of the everyday life of any child.

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One of the main aims of physical therapy is to allow children to lead a normal life and to consider themselves no different from their friends. The general advice is to let patients play the sports they want, but to instruct them to stop if muscle soreness is present. This will enable the child to start early in the treatment of their disease; partially restricted sports activities are better than being excluded from exercise and playing sports with friends because of disease. The general attitude should be to encourage the child to be independent within the limits imposed by the disease. Exercise should be undertaken after advice from a physical therapist (and sometimes requires the supervision of a physical therapist). The physical therapist will be able to advise which exercises or sports are safe, as this will depend on how weak the muscles are. The workload should gradually increase to strengthen the muscles and improve stamina.

### **3.4 Can my child attend school regularly?**

School for children is similar to work for adults: it is a place where children learn how to be independent and self-reliant as an individual. Parents and teachers need to be flexible in order to allow children to participate in school activities in as normal a way as possible. This will help the child to be as successful as possible academically, as well as helping them to integrate and be accepted by both their peers and adults. It is extremely important that children attend school regularly. There are a few factors that may cause problems: difficulty in walking, fatigue, pain or stiffness. It is important to explain to the teachers what the child's needs are: help because of difficulty in writing, proper tables to work on, being allowed to move regularly to avoid muscle stiffness, and help in participating in some of the physical education activities. Patients should be encouraged to take part whenever possible in PE (physical education) lessons.

### **3.5 Can diet help my child?**

There is no evidence that diet can influence the disease process, but a normal balanced diet is recommended. A healthy, well-balanced diet with protein, calcium and vitamins is recommended for all growing children. Overeating should be avoided in patients taking corticosteroids, as these cause increased appetite which can easily lead

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to excessive weight gain.

### **3.6 Can climate influence the course of my child's disease?**

Current research is looking at the relationship between UV-radiation and JDM.

### **3.7 Can my child be vaccinated or have immunisations?**

Immunisations should be discussed with your doctor, who will decide which vaccines are safe and advisable for your child. Many vaccinations are recommended: tetanus, poliomyelitis by injection, diphtheria, pneumococcus and influenza by injection. These are non-live composite vaccines which are safe for patients on immunosuppressive drugs. However, live attenuated vaccines are generally avoided because of the hypothetical risk of inducing infection in patients receiving high dose immunosuppressive drugs or biologic agents (such as mumps, measles, rubella, BCG, yellow fever).

### **3.8 Are there problems associated with sex, pregnancy or birth control?**

JDM has not been shown to affect sex or pregnancy. However, many of the medications used to control the disease can have adverse effects on a foetus. Sexually active patients are advised to use safe methods of birth control, and to discuss issues of contraception and pregnancy (especially before they try to conceive) with their doctor.