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JUVENILE DERMATOMYOSITIS

What kind of disease is it?
Juvenile dermatomyositis (JDM) belongs to a group of diseases called autoimmune diseases. In autoimmune diseases an abnormal reaction of the immune system causes an inflammation in body tissues when no infection is present. In dermatomyositis, the inflammation is in very small blood vessels in muscle (myositis) and skin (dermatitis). This leads to characteristic symptoms, such as muscle weakness, or pain, mainly in the muscles surrounding the hip and shoulder girdle, and skin eruptions in the face, above the eyelids, on the knuckles, knees and elbows.
The disease can be present in children and in adults. If the symptoms of dermatomyositis present before the age of 16 years, the disease is addressed as the juvenile form.

How common is it?
JDM is a rare disease in children. The incidence of JDM is estimated to be around four in 100,000 children. Girls are affected twice as often as boys. Onset is most common between the ages of four and 10 years. There is very little evidence for any geographically or racially linked predisposition to JDM.
Dermatomyositis is also seen in adults, but the presentation and course of the disease differs from the juvenile form of dermatomyositis. Unlike in adults, there is no association with the development of malignancy.

What are the causes of the disease and is it inherited?
As in most autoimmune diseases, the exact cause of dermatomyositis is not yet known. The origin of the disease is probably multifactorial, which means that a combination of genetic and environmental factors leads to an increased susceptibility to dermatomyositis. JDM, therefore, is not an inherited disease. At most, there is an increased frequency of autoimmune disease in families of children with JDM.
As for the environmental factors associated with the development of JDM, a lot of investigations have been performed. It is hypothesized that micro-organisms might trigger an abnormal response within the immune system in auto-immune diseases.

Can it be prevented?
Because a causative factor is not yet known, recommendation for prevention of the disease can not be made.

Is it contagious?
JDM is not contagious, because it is caused by an inflammation of the immune system of the patient in the absence of any infection.

What are the main symptoms?

Increasing fatigue with progressive limitation of physical fitness and mobility are usually the most prominent symptoms of muscle weakness caused by inflammation in JDM.
Muscle and joint pain can be a prominent feature and inflammation in joints is present in some children. Skin disease may either precede or follow muscle involvement. This skin disease consists of a red, often scaly, rash typically located over the top of the joints (Gottron's patches).
On the face, redness with some swelling around the eyes (periorbital erythema) and on the cheeks (malar rash) are common, as is purplish discoloration of the upper eyelids (heliotrope). The rash is often worse after sun exposure (photosensitive). The rash may be more generalised, covering other parts of the body, and ulceration can occur. Superficial vessel changes can be visible as red dots at the edge of nailfolds and eyelids.
Characteristically, muscle groups close to the trunk (proximal muscles) are involved symmetrically, often together with abdominal, back and neck muscles. In practical terms, this means that the child might start to refuse walking to school and doing sports. Small children may become fussy, requiring to be carried around. With disease progression, climbing stairs and getting out of bed might become a problem. Inflamed muscles tend to shorten (contracture) and extremities might get fixed in a bent position, with important functional consequences.
In longstanding disease, calcium might get deposited under the skin, forming hard nodules that might become ulcerated and milky liquid can drain out (calcinosis). In the most severe disease, virtually all muscles attached to the skeleton (skeletal muscles) may become affected, including those involved in breathing, swallowing and articulation. Therefore, voice changes, difficulties with feeding or swallowing, coughing and shortness of breath are important alerting signs.
Problems with the gastrointestinal tract including abdominal pain, discomfort and constipation are also common. In rare cases, occlusion of blood vessels supplying the bowels may cause severe abdominal problems.

Is the disease the same in every child?
The disease is very variable. There is a wide range, from mild disease that has minimal functional impact, to the severe, disabling condition. The organ involvement differs from child to child. There are cases with just skin disease with minimal, or absent, muscle weakness, cases of muscle disease alone (juvenile polymyositis) and profound disease affecting the skin, muscles, lung and gut.

Is it different in children compared to adults?
In adults, dermatomyositis can be secondary to underlying malignancies, but this is not the case in children. Isolated muscle involvement, without skin disease (polymyositis), is more frequent in adults, while it is very rare in children. Adults can also have positive blood tests that are rarely identified in children, suggesting differences in the underlying disease.

How is it diagnosed? What are the tests?
Diagnosis of JDM is based on the clinical features of muscle and skin involvement, described above, in combination with laboratory tests. Initially, it may look like other

diseases such as SLE, JIA, vasculitis, or a congenital muscle disease, which are distinguished by different clinical and laboratory features.
The severity of muscle involvement is tested by looking at muscle strength in different parts of the body. Small blood vessel involvement can be seen in the finger nailfolds (nailfold capillaroscopy).
In the majority of cases, affected muscles become more "leaky" and substances that are usually mainly in the muscle cells leak into the blood and can be measured in laboratory tests.
The most important of these are the proteins, called muscle enzymes. It is of note that the similar enzyme spectrum may also come from the liver. This means that certain combinations of laboratory findings, together with the clinical picture, help the clinician to distinguish between the two.
Other laboratory tests can help in the diagnosis.
Antinuclear antibodies (ANA) may be positive in this disease, as well as in other autoimmune diseases.
Blood tests are commonly used for disease and treatment follow-up (see below).
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 is rarely necessary in typical disease. Muscle inflammation can also be visualised using magnetic resonance techniques (MRI).
Muscle biopsies (the removal of small pieces of muscle) are important to confirm the diagnosis, and are a very potent research tool for better understanding the principles of the disease.
Usually other tests may be performed in order to detect involvement of other organs. Electrocardiography (ECG) and heart ultrasound are useful for heart disease, chest X-ray or CT scan together with pulmonary function tests may reveal rare lung involvement. X-ray of the swallowing process using the contrast liquid detects involvement of muscles in the throat and oesophagus.

What is the importance of the tests?
In typical cases of full blown proximal muscle weakness (involvement of muscles in the thigh and upper arm) and characteristic skin lesions, dermatomyositis, the diagnosis can be made on appearance alone. Tests are then used to confirm this diagnosis and to monitor treatment.
Standardized scores and blood tests (reflecting muscle damage). Are used to assess muscle involvement.

Therapy
JDM is a treatable disease, with medication aimed at controlling the disease process until it goes into remission. The treatment is tailored to the needs of the individual child.
If the disease is not controlled, damage can occur and this can be irreversible. This damage can produce long-term problems, including disability, which persist even when the disease has gone.
In many children, physiotherapy, and psychological support can also be important elements in the treatment of JDM.

What are the treatments?

Corticosteroids: These drugs are extremely good at controlling inflammation, wherever it is in the body. If they need to work very fast they can, especially if given into a vein. In fact, they work faster than any of the other drugs and can be life-saving. Unfortunately, they do have side effects, which is why doctors try to control the inflammation using other medication in the long-term. The side effects include stunted growth, increased risk of infection, high blood pressure and osteoporosis (thinning of the bones). All of these side effects are dose dependant, meaning that they cause few problems at a low dose, but increasing problems with increasing doses. Steroids suppress the body's own steroids and this can cause a serious problems, which can be fatal if they are suddenly stopped. They need to be reduced slowly.

Treatment with steroids is often associated with other drugs, such as methotrexate, or cyclosporine, that help in maintaining remission when reducing the steroid dose.

Methotrexate: This is a drug that takes six to eight weeks to start work and is usually given over a long period of time. Its main side effect is nausea, in addition to mouth ulcers, mild thinning of the hair and liver problems. The liver problems are mild, but can be made much worse by alcohol. It has serious effects on a developing foetus, and cannot be taken when pregnant. There is, theoretically, an increased risk of infection, though in practice, problems have been seen mostly with chicken pox.

Cyclosporin: Like methotrexate, it is usually given over a long period of time. Its long-term side effects include raised blood pressure, increased body hair, gum enlargement, and kidney problems.

Other therapeutic approaches include:

Intravenous Immunoglobulin (IVIG). This contains human antibodies concentrated from blood. It is given into a vein and works through effects on the immune system, causing an improvement in inflammation. The exact mechanism for how it works is, as yet, unknown.

In resistant disease other drugs, such as azathioprine or, in most severe cases, cyclophosphamide, may be necessary. The use of more recent drugs, such as biological agents, is still experimental in JDM. As in other systemic autoimmune diseases, it is hoped that they could represent a substantial improvement in the treatment of JDM.

Physiotherapy:

Common physical symptoms of JDM are muscle weakness and joint stiffness, resulting in a reduction in mobility and fitness. These disabilities can be helped through regular physiotherapy sessions. The physiotherapist will teach both children and parents a series of appropriate stretching, strengthening and fitness exercises. These exercises are designed 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 so that they can ensure compliance with the exercise programme.

How long should treatment last for?

The length of drug treatment will depend on the characteristics of the disease in the individual child. For some children the disease is short-lived, whilst others have the disease for many years.

Doctors aim to control the disease and treatment is only stopped after the child has been disease free for some time. JDM is a disease particularly sensitive to drug treatment reductions. Meaning, if the drugs are reduced too fast, it can cause a flare of the disease.

What about unconventional and complementary therapies?
Many unconventional therapies are proposed to patients nowadays and one has to think carefully about unqualified medical advice and its implications. If you want to take unconventional therapy, please tell your paediatric rheumatologist. Most physicians will not be opposed, provided you follow medical advice. When drugs, such as glucocorticosteroids, are needed to keep JDM under control, it is very dangerous to stop taking them if the disease is still active.

CHECK-UPS
Regular check ups to monitor disease activity and side effects of treatment are extremely important. An objective measurement of muscle power will allow monitoring of muscle weakness. As JDM can affect all parts of the body, the doctor will need to examine the whole child carefully. Controls include assessment of muscle strength and blood tests, such as muscle enzymes, and tests necessary to evaluate drug toxicity.

Prognosis
If the disease is controllable, the overall prognosis of JDM is favorable. In contrast to adults with DM, JDM is not associated with the occurrence of a malignancy. However, there is a mortality risk in those rare cases where respiratory, cardiac, neurological, or gastrointestinal complications develop during the acute phase of the disease. Functional outcome is largely determined by the development and extent of calcium deposits (called calcinosis) and the severity of muscle involvement, which can lead to muscle atrophy and contracture. Calcinosis is said to occur in 10 to 30% of all JDM children. There is no proven therapy for calcinosis.
The course of the disease can be divided into several subtypes. JDM, with a monocylic course, is defined as just one episode of disease that is in remission within two years of onset, without relapses. This form has the most favorable prognosis.
JDM, with a chronic polycyclic course, is characterized by prolonged remission with one or more relapses after stopping treatment.
Chronic, active disease, is characterized by a chronically persistent disease activity despite treatment (chronic remittent disease course). This last group has a higher risk of complications.