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Juvenile Dermatomyositis

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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 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

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 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 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 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.