Rigid spine syndrome (RSS)
(Congenital muscular dystrophy with rigidity of the spine, including RSMD1)

What is RSMD1?
The congenital muscular dystrophies are a group of conditions which share early presentation and a similar appearance of the muscle. Congenital means ‘from birth’ and in congenital muscular dystrophy the initial symptoms are present at birth or in the first few months. Congenital muscular dystrophies are a very heterogeneous group of conditions and in the last few years a lot of effort has gone into identifying the separate entities and in locating the gene responsible for a number of these forms.

The gene responsible for most cases of rigid spine syndrome (RSS) has recently been identified and lies on chromosome 1 (1p35-p36). This gene is responsible for the production of a protein called selenoprotein N (SEPN1), a protein of unknown function. The forms of rigid spine syndrome with mutations in this gene have been called RSMD1 congenital muscular dystrophy.

RSMD1 congenital muscular dystrophy has specific features such as:
- rigidity (stiffness) of the spine
- muscle weakness, which is relatively mild compared to other forms of congenital muscular dystrophy and mainly involves the muscles in the neck and trunk and, to a lesser extent, in the limbs
- respiratory problems, which often require night time ventilatory support at night

Which are the first signs?
Babies with RSMD1 often have hypotonia (low muscle tone, floppiness), and may have difficulty in holding their head or have a delay in learning new motor skills such as sitting unaided, crawling or walking in some cases.

Is RSMD1 inherited?
Yes. The pattern of inheritance is known as ‘autosomal recessive’. This means that both parents are carriers of the condition (although clinically unaffected) and they have risk of 25%, or 1 in 4, in each pregnancy of passing the condition on to their children.

How is RSMD1 congenital muscular dystrophy diagnosed?
The diagnosis of RSMD1 congenital muscular dystrophy is usually suspected from the history and examination. The next step towards the diagnosis however is generally made by looking at a piece of muscle (muscle biopsy). This involves taking out a small piece of muscle, usually from the thigh. When the muscle is studied under the microscope, it is possible to look for changes, which might indicate a muscle problem. In children with a muscular dystrophy the muscle fibres, instead of being evenly sized, show a great variation and some of these fibres are replaced by fat and fibrous tissue.

Before doing a muscle biopsy a few other tests may be done. One of these tests is a blood test, which measures the level of a muscle protein (creatine kinase or CK) that however is generally normal or only mildly raised.

Muscle ultrasound may also help to detect abnormalities in the muscle. The technique is very simple, similar to the ultrasound studies carried out in pregnancy and may provide further evidence of the involvement of the muscle. Other imaging techniques of muscle such as muscle magnetic resonance imaging (MRI) also exist and some time their use can also help the diagnosis.
These tests provide a broad indication that there is a muscle problem but cannot pin-point the precise diagnosis.

In the last few years **genetic tests** looking for abnormalities in the gene responsible for RSMD1 congenital muscular dystrophy have become available and these provide the ultimate diagnosis.

**Prenatal diagnosis** is one development in the diagnosis of inherited conditions. It is based on the ability to detect the abnormality in the developing fetus. In families who have a child with RSMD1 congenital muscular dystrophy who decide to have another baby it is possible to detect whether the baby has the same gene defect early in pregnancy.

**Is there a treatment or cure?**
At the moment there is no cure for congenital muscular dystrophy, but there are ways, described below, of helping to alleviate the effects of the condition and prevent complications.

**Can a child with RSMD1 learn to walk?**
The severity of this condition varies greatly from person to person. Almost invariably children with RSMD1 however learn to walk even though this may be delayed. Most of them maintain the ability to walk for life.

**What other physical effects might RSMD1 have on a child?**
Most of the children with RSMD1 congenital muscular dystrophy also develop a curvature of the spine (scoliosis) which should be carefully monitored.

As the muscles are weak, the child may also develop 'contractures', this means that the muscle tendons tighten up and the child is unable to move the limbs or the joints as freely as a healthy child. Physiotherapy can help prevent this and a programme of exercises should be worked out with a physiotherapist after diagnosis.

**Is RSMD 1 congenital muscular dystrophy progressive and is it life threatening?**
The condition is fairly stable and the child appears to gain strength as he or she gets older. While motor function remains relatively stable or only slowly progressive, the curvature of the spine (scoliosis) becomes rapidly more pronounced and it is important to carefully monitor the progression of the curvature and receive appropriate help (see next section).

These patients also often experience breathing problems at night because the muscles which assist breathing are affected. It is therefore very important to monitor this function on a regular basis by performing 'sleep studies'. This test is very simple and consists of applying a small wrapping around one finger. The wrapping is connected to a small machine which records the level of oxygen throughout the night. Night time breathing problems may happen in children of any age and, when present, children feel tired, often have headaches in the morning, soon after they wake up, may feel sleepy during the day and lose appetite and weight. Another problem can also be that of frequent respiratory chest infections. If these signs are present or if the level of oxygen recorded at night are not satisfactory, children can be helped by referring them to a respiratory physician who will provide a mean of supporting breathing at night (ventilator). This usually requires a special facial or nasal mask attached to a small machine that pumps air when it is needed.

Another frequent problem after the first few years is failure to thrive and it is therefore also important to monitor weight and height to be sure that children with RSMD1 congenital muscular dystrophy receive enough food and energy.
What help is available?
Physiotherapy is one of the main forms of help. An initial physiotherapy assessment at the time of the diagnosis should be followed by an exercise programme and regular check-ups. The main aim of physiotherapy is to keep the muscles as active as possible and to prevent the formation of contractures (muscle tendon tightness causing restriction in the range of joint movement). Children are encouraged to remain as active as possible. Swimming is a particular good form of exercise.

The main focus, however, has to be on scoliosis. It is essential to maintain a proper sitting and standing posture to prevent curvature of the spine, if a curvature occurs, use of a spinal brace may help to prevent further deterioration of the curvature. Surgical intervention (scoliosis surgery) might be needed in some cases.
Where can I get help?

Muscular Dystrophy Campaign
61 Southwark Street
London SE1 0HL
Tel: 020 7803 4800 (all departments)
Freephone: 0800 652 6352
Email addresses:
Information and Support Line: info@muscular-dystrophy.org
Research: research@muscular-dystrophy.org

Contact a Family
209-211 City Road,
London EC1V 1JN
Tel: 020 7608 8700
Fax: 020 7608 8701
Helpline: 0808 808 3555 or
Textphone: 0808 808 3556
Freephone for parents and families (10am-4pm, Mon-Fri)
Email: info@cafamily.org.uk
Web: www.cafamily.org.uk

Other MDC factsheets that may be useful
- Congenital muscular dystrophies
  - MDC1A (merosin-deficient congenital muscular dystrophy)
  - Rigid spine syndrome
  - Ullrich congenital muscular dystrophy
- Carrier detection tests and prenatal diagnosis of inherited neuromuscular conditions
- Inheritance and the muscular dystrophies
- Muscle biopsies
- Surgical correction of spinal deformity in muscular dystrophy and other neuromuscular disorders

MC38
Published: 12/04
Updated: 04/08
Author: MDC Research Department, in association with Francesco Muntoni, Professor of Paediatric Neurology, Hammersmith Hospital.

Disclaimer
Whilst every reasonable effort is made to ensure that the information in this document is complete, correct and up-to-date, this cannot be guaranteed and the Muscular Dystrophy Campaign shall not be liable whatsoever for any damages incurred as a result of its use. The Muscular Dystrophy Campaign does not necessarily endorse the services provided by the organisations listed in our factsheets.