For those who have been informed that the foetus has spinal dysraphism

Spinal dysraphism is a type of neural tube defect, a malformation that develops in early pregnancy and affects the brain and spinal cord. There could be a total absence of symptoms or it could lead to paralysis of the legs, reduced sensitivity and impaired functioning of the bladder and bowel. Spinal dysraphism is also known as myelomeningocele (MMC).

The symptoms depend mainly on whether the spinal dysraphism contains exposed neural tissue (open spinal dysraphism) or if it is covered by skin, and where in the spine the dysraphism occurs. Using modern ultrasound technology, other changes in the spine are sometimes detected, such as an insufficiently closed vertebral arch, which do not affect the spinal cord itself and which could mean there are no symptoms.

The description below mainly outlines the consequences of spinal dysraphism (MMC), which nowadays is normally diagnosed prenatally, i.e. during pregnancy. We describe what it is like in most cases, although in rare cases spinal dysraphism could be associated with further medical complications depending on the nature of the malformation of the brain, spinal cord and skeletal system.

When signs of spinal dysraphism are discovered in the early stages of pregnancy, the pregnant woman is offered an examination and follow-up at a regional hospital, and regular ultrasound examinations of the foetus are carried out through to delivery. Delivery is planned for when the pregnancy is considered to be full term. In most cases it will be by caesarean section at a regional hospital, where the child will then receive care. A paediatric neurologist and an obstetrician will provide all the necessary information.

Following delivery, an open spinal dysraphism is covered with moistened dressings and a sterile sheet and the new-born child, who is usually in good health, can lie with the parents. The child is then cared for at a paediatric clinic, either in the neonatal department or in the paediatric department.

Shortly after the birth, the child and the parents will meet a team of specialists, made up of a paediatric neurologist, a neurosurgeon, a paediatric orthopaedic surgeon, a physiotherapist, a urologist and a urotherapist. The team will investigate what type of treatment will be necessary and they will maintain contact with the children and young persons' habilitation department in the family's home town. The period of care required after the child is born can vary, although expect it to be around four weeks.

An open spinal dysraphism is operated on by a neurosurgeon during the first or second day of life to achieve a covering of skin. The majority of children experience hydrocephalus (a build-up of fluid in the ventricles in the brain). In that case, they will undergo an ultrasound examination, and often an MRI, and the neurosurgeon will insert a thin shunt catheter to channel the fluid from the brain to the abdominal cavity. When the child grows up, the shunt valve and catheter under the skin will be barely noticeable. For different reasons, the shunt sometimes does not work as it should and a further operation could be necessary to fit a new shunt. This may need to be done several times during the person's lifetime.

If the dysraphism is a long way down the spine, the child could have both good sensitivity and good leg movement. If it is higher up, the child will in most cases have reduced sensitivity, muscle weakness and malpositioning of the hips, knees and feet from birth. A paediatric

orthopaedic surgeon, together with a physiotherapist, will conduct an examination and recommend a series of measures that will help the child as far as possible to move around unaided. Many children learn to walk although some need splints (orthoses) and assistive aids. The majority eventually use a wheelchair.

Almost everyone with spinal dysraphism – even those who have good motor skills and can walk without assistance – experience bladder problems. This means that they could find it difficult to empty their bladder or that there is continuous leakage. In most cases the parents, just a few days after the child is born, learn to empty the child's bladder using a catheter (clean intermittent catheterisation, CIC). From school age, most children learn to manage CIC themselves. The majority will eventually need to empty their bowel with the aid of bowel irrigation, which is something they can also learn to do themselves. Regular emptying reduces the risk of leakage.

A paediatric follow-up and an orthopaedic follow-up of the spine are carried out as the child grows up. Several operations could be necessary, performed not only by an orthopaedic surgeon (due to joint malpositioning or scoliosis), but also by a neurosurgeon (in the event of shunt failure or release of a tethered spinal cord) or a paediatric urologist (to facilitate emptying of the bladder and bowel).

People with spinal dysraphism often experience difficulty with what are termed executive functions (taking initiative, focusing, organising and planning), which could impact on their day-to-day life and learning ability. Children may need help at preschool and school, both from an assistant and a remedial teacher. Some suffer from intellectual impairment. Autism could occur.

The children and young persons' habilitation unit has teams made up of different professionals. They carry out procedures, including examinations, treatment and testing of assistive aids. The procedures are based on the needs and conditions of the child and they also provide information about the various forms of support available in the community.

In summary, it can be said that people with spinal dysraphism reveal symptoms that can vary considerably in degree and it is very difficult to predict how each individual will develop. A person with spinal dysraphism needs a great deal of specialist care from the habilitation department as well as day-to-day support, although many individuals manage to live an independent life.

In Sweden, there is a national follow-up programme, MMCUP, linked to a quality register, and people with spinal dysraphism (myelomeningocele, MMC) are invited to join.

The interest groups RBU and Spin-Off can provide further information. Links to their websites can be found on the MMCUP website mmcup.se, which also contains information about the national follow-up programme. Spinal dysraphism is included at a number of the 'Centres for rare diagnoses', which are linked to regional hospitals in Sweden and are currently at the build-up stage.