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Surgical treatment of Duchenne muscular dystrophy patients in Germany: the present situation

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In 1988, we familiarised ourselves at Poitiers with the concept of operative treatment of the lower limbs and the spine in Duchenne muscular dystrophy (DMD) patients which Yves Rideau and his collaborators (1, 2) had developed there in the early 1980s. Thereupon, we immediately established the techniques at our home universities, first at the Technische Universität Aachen and, from 1999 on, at the Universitätsklinikum Erlangen, Germany. Since then, we have applied the technique to more than 500 DMD patients in total by performing more than 800 operations on the lower limbs and/or spine. In support of findings reported by Professor Rideau in this issue (3) we observed that, where patients are still ambulatory at the time of operation, the operation delays the point at which patients become wheelchair-bound by about two years. Likewise, patients receiving this treatment were/are also able to perform the Gowers' manoeuvre for around two years longer (4-6).

Key words: Duchenne muscular dystrophy, prophylactic surgery, prevention of scoliosis

The natural course of Duchenne muscular dystrophy

The disease follows a more or less regular course – though certainly with individual variations. DMD boys appear "healthy" at birth. At 3-5 years of age the first signs manifest themselves as clumsiness which is usually not recognised as being related to a muscular dystrophy. If not correctly diagnosed and treated, the boys become progressively unsteady in their walking, have a propensity to fall, use the Gower's manoeuvre to stand up again and acquire a waddling gait. Around the age of 10 they lose the ability to walk and, once in a wheelchair, they regularly develop hip and knee contractures as well as foot deformities (club feet). Concomitantly severe forms of scoliosis may appear and the Cobb's angle might still increase after the boys have stopped growing. The scoliosis thus progressively compromises ventilation and usually leads to death at age 16 to 18. This so called "natural course" can today be considerably improved by sophisticated orthopaedic surgical procedures.

Our decisive experience is that during the early stages of the disease, restricted mobility of the joints of the lower extremities may be apparent when subtle physical examination techniques are applied. Curiously, it is not so much the manifest contractures of the flexors that are relevant at the early stages of the disease, but the loss of physiologic hyperextensability of the hip, knee and ankle joints, as well as restricted leg adduction at the hip joints. This is caused by a shortening of the iliotibial tract resulting from the myopathic alterations in the *m. tensor fasciae latae* as well as by a cicatrisation of the *septum intermusculare*. This latter change forces the boy to adopt a spraddle-legged gait in order to keep the trunk upright. This, however, increases instability of his gait and the tendency to fall.

In 80% of cases these restrictions on mobility are already present at 4-6 years of age. Since the dystrophic process will still progress for a long time thereafter, we recommend this to be the ideal age for afflicted boys to undergo prophylactic surgery to prevent contractures if they:

- show this early restriction of joint mobility,
- have a total muscle force of > 70% (force of quadriceps femoris at least 3 MRC), and
- are able to rise from a supine position to standing in less than 5 seconds (7).

Actions required for prophylactic surgery

The first stage of this prophylactic operation involves severing the tendons of the spinal muscles (*mm. tensor fasciae latae, sartorius and rectus femoris*) and mak-

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ing a proximal incision in the iliotibial tract that reaches down to the gluteal fascia. Next comes the excision of the iliotibial tract with the attached *septum intermusculare*, combined with a transection of the fibrotic fibres of the *m. biceps femoris*. This is done by lengthening the Achilles tendon from the front with a Z-shaped diagonal cut and then sewing it back together, allowing some slack. The last part of the surgical procedure is a subcutaneous medial tenotomy of the knee flexor tendons. Both legs are operated on in the same session. No plaster is used. If subtle haemostasis is achieved Redon drainage is not necessary. Absorbable suture material is used for the threads so there are no stitches to remove. Thus overall patient morbidity is very low.

Anaesthesia of the patients is without triggering substances (relinquishment of volatile anaesthetics and succinylcholin). To reduce postoperative pain and to administer intraoperative analgesics a sacral catheter is inserted (8-12).

When should prophylactic surgery best be performed?

If the boys receive surgery at the optimum time (i.e. before loss of ambulation) they regain their ability to stand 2 days after the operation; walking is possible, as a general rule, after 4 to 6 days (4-7). If a boy first presents at a later age, say shortly before he loses his ability to walk, or indeed just after he has lost it, we still recommend that his lower limbs are operated on to treat hip, knee and ankle contractures. The aim of this treatment for a wheelchair-bound patient would be to enhance the likelihood of being able to stand with the help of ortheses or to use a chair with prop-up facilities. Daily exercise requiring standing periods of up to 2 hrs has been shown to slow the progression of the inevitable scoliosis and the ensuing impaired respiration (13).

When a patient has completely lost ambulation the operative regime has to be adapted according to the status of the contractures. Soft-tissue operations on hip and knee contractures exceeding 30° are of limited benefit. However, they may prevent increased severity of contractures. Surgical treatment of the feet is particularly important in patients with increasing talipes. If such feet remain untreated, the patient will soon be unable to place them in the orthograde position on the footrest of the wheelchair. Even if treated with orthopaedic shoes the progression of the deformities and the patients' discomfort cannot be improved. We cannot confirm the observations by Leitch et al. (14) that operative treatment has no advantage over conservative treatment. We perform on wheelchair-bound patients a rather complex form of operation by not only severing of the Achilles' tendons, but by also resecting each end by about 2 cm so that they cannot grow together any more. In addition we use the same technique for the tendons of *mm. tibialis posterior, flexor digitorum longus and flexor hallucis longus* (7). The substantial majority of patients receiving this treatment do not need orthopaedic shoes.

Do other forms of symptomatic treatment interfere with prophylactic surgery?

The well established treatment of DMD patients with prednisolone or deflazacort results in increased total muscle strength and has side effects that can usually be managed (15). With these patients we observe fewer indications for prophylactic treatment against contractures in the lower limbs. With appropriate perioperative substitution of the medication we found no negative influence on the feasibility and the success of these operations when patients had undergone steroid medication.

These days when a wheelchair-bound patient develops a progressive scoliosis (> 20°) we recommend surgical stabilisation of the spinal column from T2 or T3 to S1 because this is the best way to improve his sitting position. We do not consider a corset as an alternative unless surgery is not appropriate for the patient or he refuses it.

Meanwhile use of assisted ventilation has become widespread and we now often see patients equipped with a small respirator even before they have had operations on their spinal column. In our experience such patients with unequivocally reduced pulmonary function can be safely operated on so that their sitting stability – an important contribution to their quality of life – is conserved.

Additional reasons in favour of surgical treatment

In spite of the time window mentioned above for optimal operating conditions we would like to stress that surgical stabilisation of the spine should nevertheless be aimed at, because the feasibility of correction to the spine and the associated improvement of pulmonary function are better, the smaller Cobb's angle is (1). Moreover the operation is better tolerated and the younger a patient is, the easier it is to manage any perioperative complications (16).

Technical details of the operation

Operations on the spine should only be performed in hospitals with maximum medical care, where surgeons are well versed in the aforementioned techniques, and are competent in the relevant anaesthesiologic and haemostasiologic aspects of intensive care (17, 18). For the stabilisation of the spine we prefer instrumentations (e.g. ISOLA systems, XIA, CDH legacy), that allow us to use pedicle screws along the lumbar spine and, as a rule, also in the sacral bone. For the thoracic vertebrae we apply an

instrumentation using clamps with hooks around the rib processes and laminae of T3 to T5 and Luque wires from T6 to T12 (19). This results in a semi-rigid construction that has so far not shown a fatigue failure of the instrumentation in any of our more than 300 patients.

In our experience it is essential to include the sacral bone in the instrumentation because this is the only way to obtain an efficient correction of an oblique pelvis. Other teams, however, have different opinions on this point (16). Perhaps this is because they have treated scoliosis with significantly lower Cobb angles than we have.

Conclusion

We find that patients, who are treated according to this comprehensive concept, i.e. receive complex medication, undergo the sophisticated surgical programme detailed above and accept assisted ventilation, may have a life expectancy of over 30 years with a quality of life that patients at the age of 10 years could never have hoped for thirty years ago.

To establish whether or not surgery is indicated in patients with any muscular dystrophy, three key questions should be considered:

1. Will surgical treatment really improve the patient's quality of life, i.e. does it advance his individual psycho-social situation? On the other hand, one may ask whether there are psychological or physical reasons why the patient and his family should be spared such serious intervention.
2. Which of the various operative *techniques* is the best choice for a case in question?
3. What is the best point in time for a prophylactic operation?

As mentioned above, the burden of operations on the lower limbs and the spine is smaller when these are carried out at an early age because less invasive techniques can be used, and the patient's condition is better.

The decisive criterion for optimal orthopaedic treatment of a patient with degenerative muscle disease is his early presentation to an experienced orthopaedic team so that the full prophylactic concept involving conservative and surgical measures on limbs and spine can be applied. The most important factor in striving for long life of high quality is to conserve the patient's ability to stand for as long as possible, even if this is achieved by adjuvant appliances such as ortheses. An erect stance has many benefits for these patients, such as prevention of contractures, deformities, scoliosis and osteoporosis, as well as stimulating the circulation, promoting independence, and facilitation for attending persons.

Regular check-ups for the patients at intervals of 3-12 months – depending on the progression of the underlying disease – are desirable.

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