er, their awareness of these boundaries should not dishearten the parents and carers of PWS patients. Their input, combined with the support of a range of specialists, is vitally important for the patient's physical and mental well-being and is always worthwhile, even when the syndrome is not diagnosed immediately.

Main Forms of Treatment

The main difficulty with PWS is the sheer variety of the symptoms. Firstly, someone, such as the doctors in the maternity ward or, later on, the relevant pediatrician, needs to consider this diagnosis and have it confirmed. Usually, the case is automatically referred to a geneticist, who often explains the consequences of the diagnosis in detail to the parents. The parents then need a doctor who is familiar with the syndrome and who can explain the diversity of symptoms and create a relationship of trust. The doctor concerned should also be able to anticipate the specific problems caused by PWS and call in the various specialists at the right time in order to enlist their help. It is irrelevant which of the various specialists takes on this coordinating role. However, it is vital for the quality of life of the child concerned and his/her parents that this key function is fulfilled by a competent individual.

Restricting Calorie Intake

Controlling and restricting calorie intake was the first form of therapy that was used and remains the most effective means of weight management in PWS patients. The key to this treatment is to make sure that the patient receives the correct amount of food. Parents and teachers have to keep a close eye on all food, which is only possible if foods are strictly monitored and rigorously locked away. However, even such strict supervision can only limit and not prevent obesity.

Growth Hormone Treatment

Ever since the syndrome was discovered, short stature has been a known symptom of PWS. In the late 1980s, it was also discovered that PWS children have low muscle mass – in contrast to the muscle mass of most overweight children. Since small stature, increased fat mass and low muscle mass are all typical symptoms of growth hormone deficiency, children with PWS were first given growth hormone treatment in 1990. Several studies have shown that growth, body composition and physical performance can be significantly improved by growth hormone therapy. [fig. 4 a/b]

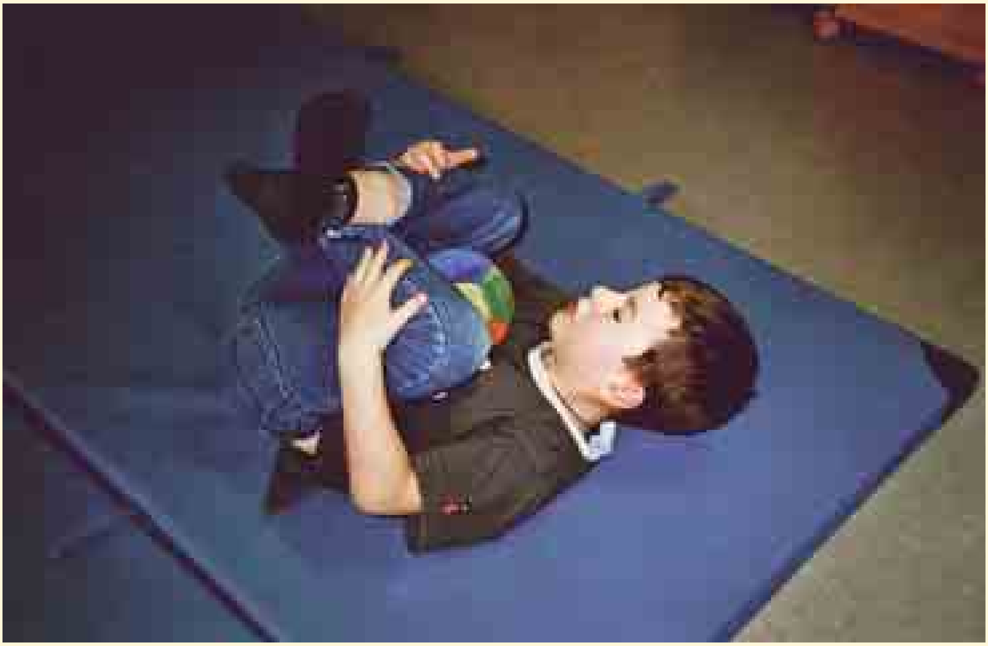
[Fig. 4 a/b] One of the first boys with PWS who was treated with growth hormone. Before treatment and after one year of therapy.

Daily Exercise

Inactivity and a lack of enthusiasm for physical exercise are also typical symptoms of PWS. Although growth hormone treatment can bring growth rates back to normal and improve body composition, the patient's muscle mass remains low and fat mass stays high, even if GH treatment is given over a long period and the patient's weight is normal. In order to improve muscle mass and increase energy consumption, it is sensible for the children and adults concerned to follow a structured daily exercise programme adapted to their own personal abilities and preferences. Parents, carers and PWS patients themselves need to be made very aware of the importance of physical activity. [fig. 5]

Sex Hormone Treatment

Incomplete pubertal development – for example, the fact that teenage boys' voices do not break – is very hard for PWS patients to cope with. The cause of this – hypogonadism (hypofunction of the testicles or ovaries) – is a central characteristic of PWS and affects not only pubertal development and psychological maturity, but also growth and body composition. Although hypogonadism is a well-documented symptom of PWS, sex hormone substitution is, to some extent, still a controversial therapy. This is related to the long-held belief that replacement with the male hormone testosterone can cause aggressive behavior in



[Fig. 5] A 6-year-old during his daily physical exercise, which helps to improve muscle mass and increase energy consumption.

young men. The effects of puberty and sex hormone substitution on the behavior of PWS patients have never been scientifically investigated. We have always achieved good results through sex hormone substitution when such therapy has been started at the right time from a physiological point of view.

Early Support, Physiotherapy, Speech Therapy

Depending on the specific needs of the child and the types of therapy available locally, the necessary support is warranted from infancy. For the reason of the permanent hypotonia and low muscle mass, physiotherapy should primarily be aimed at strengthening the muscular system. Early support is designed to encourage the child to develop. If the child's speech development is delayed or impaired, it is important to seek the advice of a speech therapist at around the time of the child's third birthday.

Psychological Consultation

Families with PWS children are usually more prone to psychosocial problems than other families. It has also been shown that the style of upbringing can have a significant impact on the weight of children with PWS. Psychological counselling for parents or families is therefore designed firstly to support the people concerned and help them use their resources wisely and, secondly, to help them bring up the child with greater clarity and consistency. However, experience shows that families often find it hard to admit that their own resources are no longer sufficient and that they need outside help and support.