Intramedullary rodding in type III osteogenesis imperfecta

Effects on neuromotor development in 10 children

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We studied retrospectively gross motor development and the impact of intramedullary rodding in 10 children with type III osteogenesis imperfecta (OI). There was a pronounced delay in motor development and the order in achieving gross motor milestones differed from the normal developmental sequence. Static milestones developed at an earlier stage than dynamic milestones.

Intramedullary rodding of the lower extremities prior to the age of 3.5 years enhanced neuromotor development, especially regarding the milestones supported standing, rolling from prone to supine and crawling with abdomen on the floor.

The different sequence in achieving gross motor milestones should have implications for future rehabilitation programs and for orthopedic surgery.

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Osteogenesis imperfecta (OI) is a congenital connective tissue disease. The incidence is about 1/5,000–30,000 individuals (Byers et al. 1992). The phenotype varies from death in the perinatal period to normal lifespan, with a minimal increase in fracture incidence (Sillence et al. 1979, Byers et al. 1988).

Major clinical characteristics of OI are osteopenia with bone fragility, variable degrees of short stature and progressive skeletal deformities. Additional clinical manifestations are blue sclerae, dentinogenesis imperfecta, joint laxity and onset of deafness at maturity (Vetter et al. 1992). Sillence et al. (1979) have identified 4 subgroups based on clinical and hereditary findings. OI type III, the most severe type compatible with life, occurs in close to one third of all individuals with OI (Vetter et al. 1992, Brenner et al. 1993). In this type, moderate deformity at birth is observed and deformation of the bones is usually progressive. The stature is very short. Sclerae are variable in hue and often lighten with age. Dentinogenesis imperfecta and hearing loss are common. Autosomal dominant inheritance is common.

OI in childhood, depending on its severity, has a large impact on neuromotor development. Rehabilitation-programs, as regards handling, positioning, physical therapy and bracing, have been designed to achieve optimal development (Binder et al. 1984, Gerber et al. 1990, Binder et al. 1993). There is no detailed study on gross motor development in young patients, or on the influence of intramedullary rodding on the progress of neuromotor development.

Several questions remain to be answered.

Is the developmental sequence in OI patients different from that in healthy children?

What is the impact of intramedullary rodding on neuromotor development, and is there an appropriate age for this intervention, in order to acquire optimal neuromotor development?

In our opinion these questions should be addressed separately for different clinical types of OI. We evaluated patients with type III OI.

Patients and methods

We studied gross motor development retrospectively in 10 children with OI type III. The mean age at follow-up was 6 (2–10) years and the mean follow-up period was 4 (2–10) years. Neuromotor development was assessed every 6 months.

The age and sequence for achieving gross motor milestones were compared between the patients and a normal population, using the van Wiechen method (1983) for screening gross and fine motor development, validated for Dutch children. Motor skills are divided into static and dynamic milestones. Normal

Table 1. Gross motor development in type III OI related to the normal population

Skill (milestone)	Type [a]	OI III Median Years	Normal van Wiechen 90th percentile Years
Lifting the head in prone position	D	3.6	0.3
Pulled by the arms from supine to a sitting position without presence of head lag	D	3.8	0.5
Rolling from supine to side-lying [b]	D	1.8	0.6
Rolling from supine to prone	D	3.8	0.7
Complete head control in sitting position	S	1.7	0.7
Sitting unsupported	S	2.1	1.0
Crawling with abdomen on the floor	D	4.8	1.0
Standing with support	S	4.8	1.0
Bottom shuffling [c]	D	4.1	–

a D dynamic, S static.
All skills according to Van Wiechen (1983), except b and c.
b Bayley Scales of Infant Development (1983).
c No reference value exists.
In OI, the results refer to the age when 6 or more of the 10 patients achieved the motor milestones.

motor development is defined as the age at which 90 percent of the population has reached a certain motor milestone (Table 1). Representative gross motor milestones were chosen that were achieved by at least 6 patients. Two motor activities observed in OI patients are not included in the van Wiechen method. One of these skills, rolling from supine to side-lying position, was derived from the Bayley Scales of Infant Development (van der Meulen and Smrkovsky 1983). Regarding the other, bottom shuffling, no reference value exists. This item was scored positive when the child was able to transfer the body in a sitting position by propulsion of the legs without use of the upper extremities. To reveal skeletal disproportions, measurements of head circumference and body length were performed.

In all children, intramedullary stabilization of the lower extremities was performed with expanding rods, as described by Bailey and Dubow (1981). The mean age at surgery was 3.5 (2–6) years. 5 children underwent surgery before the age of 3.5 years, 5 children after 3.5 years.

Results

There was a relative macrocephaly in all patients; the head circumference was between the 10th and 97th percentiles and body length was far below the 3rd percentile.

All patients had a severe delay in gross motor development. The sequence in achieving gross motor milestones was also different from normals. Static milestones, such as complete head control and sitting without support, developed earlier than dynamic milestones, such as lifting the head in prone position, rolling over from supine to prone and bottom shuffling. Rolling from supine to side-lying, developed much earlier than rolling from supine to prone (Table 1).

Gross motor milestones within 2 years of age (rolling from supine to side-lying, complete head control in sitting position, and sitting unsupported) were achieved in 9 patients before intramedullary rodding. Only 1 patient, operated after 3.5 years of age, was unable to sit unsupported until 15 months after surgical intervention.

1 patient was able to stand before intramedullary rodding. After rodding, 7 patients were able to stand with support.

Children who underwent surgery before the age of 3.5 years could stand with support sooner than children operated on later (Table 2). In other gross motor

Table 2. Months between surgery (A) and standing with support (B) in 10 patients with type III OI

Patient	A	B	A—B
1	25	20	–5
2	26	36	10
3	30	41	11
4	34	50	16
5	38	47	9
6	48	–	> 15
7	51	66	15
8	59	84	25
9	65	71	6
10	66	–	> 15

milestones, such as crawling with the abdomen on the floor and rolling from supine to prone, the same tendency was observed. Bottom shuffling was noted in 6 patients following surgery. 2 patients had this finding before rodding.

2 patients who were operated after 3.5 years of age were not able to shuffle in a sitting position. All patients who had been operated on at an earlier age successfully achieved all the motor milestones.

Discussion

Several authors have observed changes in patterns of gross motor development in children with other chronic handicaps (Adelson and Fraiberg 1974, Jaffe et al. 1988).

We found that in children with OI type III the course of gross motor development also was changed; there appears to exist a specific natural course in gross motor development in these children. There may be several reasons for this pattern.

Earlier achievement of static than dynamic milestones may be caused by the risk of fractures due to moving. It seems that the infant prevents itself from fracturing. For example, rolling from supine to prone position (dynamic milestone) is presumably avoided because of the risk of fracturing the humerus.

A delay in the craniocaudal development of muscular extension tone in a prone position, which develops in normal children in the first year of life (van Wiechen 1983), may be due to the discrepancy between head circumference and body length, with insufficient muscle strength for lifting the head in a prone position.

Shapiro (1985) stated that intramedullary rodding improved the outlook for ambulation. Porat et al. (1991) compared the function after elongating and non-elongating rodding in patients with types I, III and IV OI. They found an improved ambulation after operation. Cole (1993) observed that early surgery in type III OI reduced the incidence of fractures. Ryöppy et al. (1987) recommended early intramedullary stabilization, even in the early postnatal period. Although previous reports linked intramedullary stabilization to ambulation, we have found no detailed study on gross motor milestones in young patients with OI, or data concerning the appropriate age for surgery. In our study, the patients who were first operated on were older than 3.5 years. In the hope that earlier surgical intervention might give better results, we started to perform surgery at a younger age. Our findings indicate that intramedullary rodding, before the age of 3.5 years, yields better neuromotor development than later surgery. Because of the small number of patients and the absence of a control group, the question is whether this improvement is caused by early surgery or by selection bias.

We think that unsupported sitting is a prerequisite for intramedullary rodding. In our patients, the median age for unsupported sitting was 2.1 years. Therefore, we recommend that intramedullary rodding should be performed between the ages of 2 and 3.5 years.

We wish to emphasize that rodding does not always improve neuromotor development; not all our patients were able to achieve all gross motor milestones.

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