Ortopedia *Traumatologia* **Rehabilitacja**
© MEDSPORTPRESS, 2011; 1(6); Vol. 13, 27-36

Contractures of the Lower Extremities in Spinal Muscular Atrophy Type II. Descriptive Clinical Study with Retrospective Data Collection

Albert Fujak[1(A,B,D,E,F)], Carsten Kopschina[2(A,C,D,F)], Florian Gras[3(B,E)], Raimund Forst[1(F)], Jürgen Forst[1(A,B)]

[1] Department of Orthopaedic Surgery, Friedrich-Alexander-University Erlangen-Nuremberg, Germany
[2] Department of Trauma and Orthopaedic Surgery, Klinikum Nürnberg Süd, Germany
[3] Department of Trauma-, Hand- and Reconstructive Surgery, Friedrich-Schiller-University Jena, Erlanger Germany

SUMMARY

Background. Early development of contractures and progressive scoliosis with pelvic obliquity are the most significant orthopaedic problems for children with the intermediate form of spinal muscular atrophy, SMA type II. This study deals with the restrictions of the passive range of motion and the development of contractures of the lower extremities in these patients.

Material and methods. We followed up 143 patients, 74 female and 69 male, with SMA type II for an average of 5.3 years (±4.0, 0.2 – 18.7). Their average age at the first examination was 8.4 years (±6.6, 0.1 – 34.1) and at the last examination 12.3 years (±6.5, 0.7 – 37.1). The passive range of joint motion was determined using a goniometer. According to Johnson et al. (1992), we calculated the relative contracture index (CI).

Results. The loss of range of motion (ROM) and the contractures of the joints of the lower extremities (hip, knee and ankle) develop early and increase progressively with age. Marked restriction of motion with severe flexion contractures in some cases was observed in the knee followed by the hip and ankle joint.

Conclusion. The findings of this study give us more information about the development of contractures and deformities of the joints of the lower extremities and aim to help to improve the quality of orthopaedic care of patients with SMA type II.

BACKGROUND

The purpose of this study is to determine the development and course of contractures of the lower extremities in patients with the intermediate form of spinal muscular atrophy (SMA type II), following a survey of 143 patients.

SMA type II is a hereditary autosomal recessive neuromuscular disorder with muscular hypotonia, hyporeflexia and diffuse general weakness, most prominent in the lower extremities and proximal muscles. The distal muscles and upper extremities are also affected. Children with SMA type II are able to sit on their own but will never achieve the ability to stand or walk independently. Occasionally, they learn to stand with the support of assistive devices. The onset of SMA II usually occurs before 18 months of age [1-3].

Mostly, the clinical course is marked by stable periods of varying lengths without manifest progression of the symptoms [1,4]. In their study of 240 patients with SMA type II, Zerres et al. [1] indicate a survival rate of 98.5% for 5 years, 82.8% for 15 years and 68.5% for 25 years.

The fundamental orthopaedic problems for patients with SMA type II are contractures in the lower and upper extremities and sitting instability caused by progressive scoliosis with increasing pelvic obliquity [1-3].

Restrictions of movement and contractures appear mostly first in the lower extremities, in fewer cases they appear simultaneously in both the upper and lower extremities and in only a few patients do they appear only in the upper extremities [4-8].

To date there is no causal therapy for SMA. The course of the disease and, above all, the patient's quality of life can be greatly improved by established medical procedures. Orthopaedic treatment includes conservative methods, such as physiotherapy, orthoses and assistive devices, as well as surgery to stabilize the spine and treatment for contractures and deformities in the lower extremities [2-4]. Surgery of the upper extremities is indicated very rarely. Daily activities involving the upper extremities are made easier by the development of compensatory and adaptive techniques, which often occur spontaneously or are facilitated by simple assistive devices [3].

Surgical treatment of the contractures and deformities of the lower extremities is indicated to improve the sitting position and ability to stand with orthoses and assistive devices, to enable the fitting of shoes, to relieve pain and to enable easier care [2-4].

However, when an operation is indicated, it has to be taken into account that the strong fibrotic tendency of muscles and the biomechanically awkward sitting position in a wheelchair limit the effectiveness of such surgery, particularly of the hip and knee joints.

Among the surgical procedures which can be carried out as required in a number of combinations are the following: tenotomy or, if need be, lengthening of the sartorius, tensor fasciae latae, rectus femoris, iliopsoas, the adductors and knee flexors, and achillotenotomy. Obvious muscular imbalance and deformities of the feet could be treated by tenotomy or transposition of the tibialis posterior, flexor digitorum longus, or flexor hallucis longus muscle or the peroneal tendons and the peronaeus tertius muscle. In some cases it is necessary to carry out capsulotomy of the ankle joint in order to achieve the desired correction [3,4].

Immobilization after operative treatment of contractures or deformities of the lower extremities should be as short as possible. Longer immobilization leads to rapid deterioration of motor ability [3].

The indications and timing of surgery of the lower extremities in SMA II vary from individual to individual. Competent planning of both conservative and operative management of contractures and deformities of the lower extremities in SMA II patients requires exact knowledge and experience of the development and course of such contractures in these patients.

MATERIAL AND METHODS

This study is a retrospectively-collected data set of 143 patients with SMA type II (74 females and 69 males). These patients were examined during the regular nationwide consultations for patients with neuromuscular disorders. All patients with confirmed diagnosis of SMA with type II of SMA (patients are unable to walk but they are or were able to sit unsupported) were identified for our study. This represents the entire population of our patients. The diagnosis of SMA was confirmed by human genetics in 117 patients (82%) and by electrophysiology and muscle biopsy in 26 patients (18%).

This study includes more than one evaluation of some of the patients. Thirty-seven patients were examined once and 106 were examined a number of times, on average 6 times (±5, 2-25 times), at intervals of at least 6 months. Their average age at the first examination was 8.4 years (±6.6, 0.1-34.1 years) and at the last examination 12.3 years (±6.5, 0.7-37.1 years). The mean time of follow-up of those patients who were examined a number of times was 5.3 years (±4.0, 0.2-18.7 years).

The data are presented in a cross-sectional manner. The patients are divided into age groups: 0-2, 3-5, 6-10, 11-15, 16-20, 21-30 and over 30 years of age. Listed in the age range is only one evaluation from each patient that manifested an abnormality during that period. If a patient within one of the age groups had numerous examinations, only the examination that showed the greatest loss of range of motion was taken into consideration. It was almost always the last examination during this period.

Two experienced examiners used a goniometer to assess the passive range of motion. Results were documented with the neutral-null-method and the data was evaluated with the Microsoft Access data processor programme and Excel spreadsheets.

The neutral-null-method is based on a defined anatomically normal position, the neutral or functional position, called the neutral-null position (standing upright, with hanging arms, thumbs forward, feet close together and parallel, eyes directed forward). [9] The range of motion for every joint is described by three numbers: i.e. the range of flexion, neutral-null position (0° if achieved) and the range of extension (e.g. FL/EXT of the hip 120°/0°/10°). If the anatomically normal position (the neutral-null position) cannot be achieved, then 0° is either in the first or in the third position, depending upon the particular movement restriction present (e.g. maximal flexion of 90° and flexion contracture of 40° of the hip: FL/EXT 90°/40°/0°).

The hip was examined for flexion/extension, external/internal rotation in 90° flexion and abduction/adduction in 90° flexion. Normal physiological values were defined as [9]:

- Flexion (FL) / extension (EXT) 130°/0°/10°,
- External rotation (ER) and internal rotation (IR) in 90° flexion 40°/0°/30°,
- Abduction (ABD) / adduction (ADD) in 90° flexion 70°/0°/30°.

Flexion/extension was tested in the knee. The normal physiological values are [9]:

- Flexion (FL) / Extension (EXT) 150°/0°/5°.

The ankle was tested for dorsiflexion and plantar flexion [9]:

- Dorsiflexion (DF) / plantar flexion (PF) 30°/0°/50°.

A loss of 10° or more from the norm was considered to be a loss of range of motion.

Particularly manifest flexion contractures of the hip, knee and ankle joint (equinus) were evaluated separately. A loss of range of motion of 5° or more than the neutral-null position was considered to be a manifest contracture.

We took 10° of hip extension and 5° of knee extension as full extension – overextension.

We calculated a relative contracture index (CI) according to Johnson et al. [6]. We defined the relative contracture index as the proportion of patients with a loss of range of motion (or a manifest flexion contracture of the hip, knee and ankle joint contracture (equinus)) expressed as a percentage, multiplied by the mean maximum loss of range of motion in degrees and divided by 1,000 [6]:

$$\frac{\text{(Percentage of subjects with loss of ROM) x (Mean maximal loss of ROM in degrees)}}{1{,}000}$$

This formula results in a CI of 0 to 15, according to the number of examinations and the extent of the loss of range of motion. For example: 100% of patients with contractures and a mean maximum loss of range of motion 100°, divided by 1,000 results in a CI of 10.0.

The higher the percentage of patients with loss of ROM and the cross-sectional assessment of the mean maximal loss of ROM, the greater is the potential for contracture development. [6] According to Johnson et al. [6], the risk of developing a range of motion limitation or a contracture is high when the CI is more than 1.0, moderate when it is 0.5 to 0.9 and mild when it is lower than 0.5.

The CI of the right and left joints of the lower extremities of each patient was calculated separately and the results were displayed in diagrams for each age group. Since on average the results for right and left were comparable, they have been combined in the tables showing the loss of range of motion.

This study complies with the ethical standards for human research and has been approved by the Ethics Committee of Friedrich-Alexander-University Erlangen-Nuremberg.

RESULTS

Hip joint

A total of 396 examinations of hip joints (both right and left) were evaluated. Loss of range of motion in flexion/extension was observed in 379 cases (96%) (Table 1). This loss of ROM and the CI increased rapidly up to age 16-20 (Table 1 and Diagram 1). In older age groups, there is insufficient data for a reliable assessment. Even in 98% of the 6- to 10-year-olds (19 examinations) the ROM in FL/EX was restricted on average to 48° (±21°).

Tab. 1. Loss of range of motion (ROM) of hip joint in SMA Type II. Combined data for right and left limbs

Patients' age (years)	Number of examinations	Loss of ROM present (%)	No loss of ROM (%)	Loss of ROM [°]		
				Median	Mean (s_D)	Min - Max
FL/EXT						
0 - 2	12	7 (58%)	5 (42%)	20°	20° (10°)	10° - 40°
3 - 5	70	62 (89%)	8 (11%)	30°	31.5° (18°)	5° - 80°
6 - 10	195	191 (98%)	4 (2%)	50°	48° (21°)	5° - 95°
11 - 15	93	93 (100%)	0 (0%)	65°	67° (20°)	10° - 135
16 - 20	15	15 (100%)	0 (0%)	85°	81° (21°)	40° - 135°
21 - 30	6	6 (100%)	0 (0%)	72°	77° (14°)	60° - 100°
> 30	5	5 (100%)	0 (0%)	60°	60° (19°)	30° - 90°
All examinations	396	379 (96%)	17 (4%)	50°	55° (24°)	5° - 135°
ER/IR						
0 - 2	12	2 (17%)	10 (83%)		10° and 25°	
3 - 5	70	10 (14%)	60 (86%)	10°	16° (8°)	10° - 30°
6 - 10	195	26 (13%)	169 (87%)	10°	12° (7°)	5° - 40°
11 - 15	93	35 (38%)	58 (62%)	12.5°	16° (7°)	5° - 30°
16 - 20	15	6 (40%)	9 (60%)	20°	23° (12°)	10° - 50°
21 - 30	6	5 (83%)	1 (17%)	10°	10° (3°)	5° - 15°
> 30	5	1 (20%)	4 (80%)		10°	
All examinations	396	85 (21%)	311 (79%)	10°	15° (8°)	5° - 50°
ABD/ADD						
0 - 2	12	7 (58%)	5 (42%)	20°	22° (14°)	5° - 45°
3 - 5	70	49 (70%)	21 (30%)	20°	26° (17°)	5° - 65°
6 - 10	195	145 (74%)	50 (26%)	40°	36° (17°)	5° - 90°
11 - 15	93	80 (86%)	13 (14%)	50°	48° (16°)	5° - 90°
16 - 20	15	13 (87%)	2 (13%)	50°	50° (18°)	10° - 80°
21 - 30	6	5 (83%)	1 (17%)	57.5°	59° (12°)	45° - 80°
> 30	5	3 (60%)	2 (40%)	50°	50° (14°)	30° - 70°
All examinations	396	302 (76%)	94 (24%)	40°	41° (19°)	5° - 90°

Flexion continued to decrease until the ages of 21 to 30 years and at the same time the flexion contracture increased up to an average of 45° in the 16- to 20-year-olds (Table 4 and Diagram 4). The development of flexion contractures showed steady progression, so that there was already a contracture in 94% of examinations of those aged 11 to 15 (Table 4 and Diagram 4).

Loss of physiological overextension of the hip joint was observed in 360 of 396 cases (91%) (Table 5).

On average, there was a mobility restriction of 15° (± 8°) in external or internal rotation (Table 1) but no manifest rotation contractures were detected.

Altogether, restriction of range of motion of abduction/adduction was observed in 302 cases (76%). Here, too, there was a continuous increase in the CI for loss of ROM up to the 21-30 age group (Table 1 and Diagram 1). One patient among the 6- to 10-year-olds had a manifest abduction contracture of 5° and one patient in the 16-20 age group had a manifest adduction contracture also of 5°. The mean value for abduction and adduction shows a continuous decrease in abduction ability. There were only slight changes in adduction with age.

Knee joint

We found a loss of ROM in the knee joint in 399 of a total of 408 examinations (98%). The mean loss of ROM was 66° (± 29°) (Table 2). A mean loss of ROM of 44° (± 24°) appeared already in 97% of examinations in the 3- to 5-year-old group.

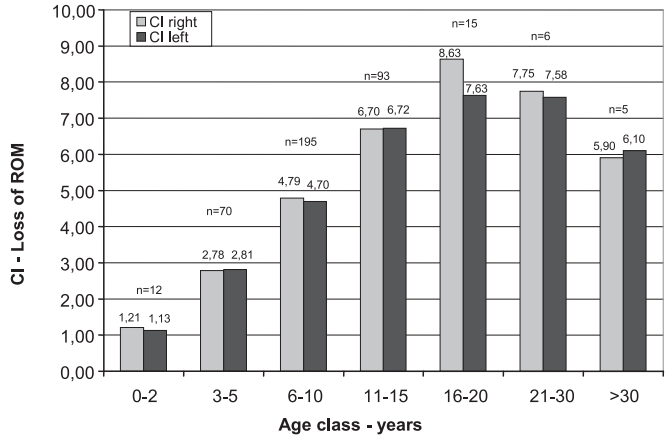
The CI showed a steady increase with values up to 9.2, which is in keeping with the extreme restrictions of mobility (Diagram 2). Manifest flexion contractures of the knee joint were found in 378 examinations (93%) and were on average 37° (± 24°) (Table 4). These contractures increased steadily into adulthood (Table 4).

Flexion of the knee joint initially remained unchanged and later seemed to show a tendency to decrease after the ages of 21 to 30. However, only limited assessment is possible because of the low numbers of examinations in the 21-30 age group and those older than 30 years.

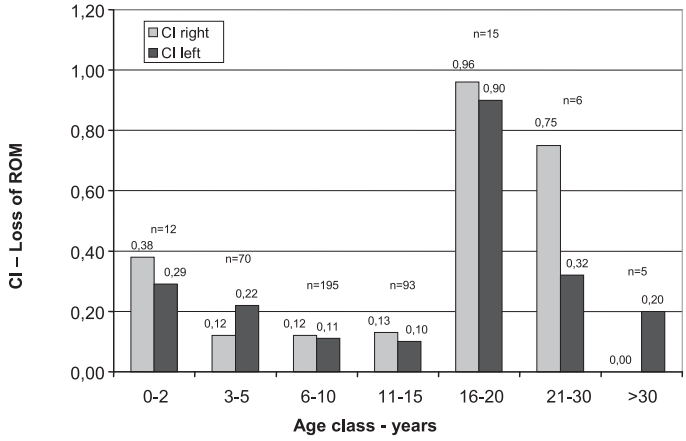
There was overextension of the knee joint in 3% of all the examinations of patients up to the age of 6 to 10 but it had completely disappeared after the age of 15 (Table 5).

Ankle joint

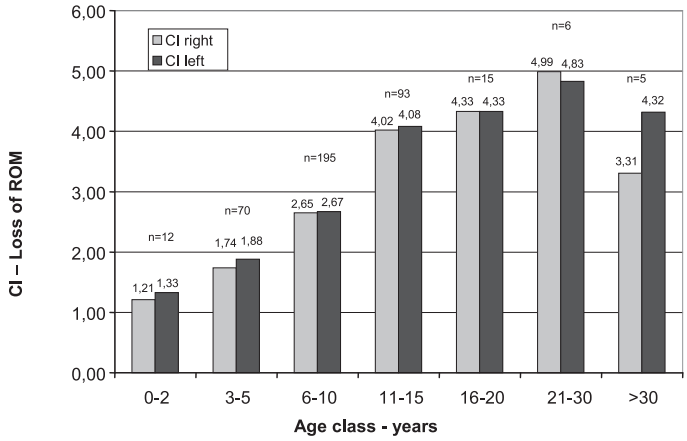
In 95% of 381 examinations of ankle joints there was restriction of ROM of 55° (±13°) on average (Table 3). As early as 3-5 years old, the average ROM in 91% of the examinations was 53° (± 11°) and the CI at this age was 4.7 (Diagram 3). Flexion contractures of the ankle (= equinus) appeared in 120 of 381 examinations (31%), with the mean value at



FL/EXT



ER/IR



ABD/ADD

Fig. 1. Loss of range of motion (ROM) – relative contracture index (CI) – hip joint FL/EXT, ER/IR and ABD/ADD in SMA Type II

Tab. 2. Loss of range of motion (ROM) of knee joint FL/EXT in SMA Type II. Combined data for right and left limbs (Loss of normal physiological ROM – with physiological extension – 150°/0°/5°)

Patients' age (years)	Number of examinations	Loss of ROM present (%)	No loss of ROM (%)	Loss of ROM [°]		
				Median	Mean (s_D)	Min - Max
0 - 2	12	8 (67%)	4 (33%)	30°	32° (14°)	10° - 60°
3 - 5	70	68 (97%)	2 (3%)	40°	44° (24°)	5° - 130°
6 - 10	197	194 (98%)	3 (2%)	55°	60° (28°)	10° - 140°
11 - 15	100	100 (100%)	0 (0%)	77.5°	75° (28°)	20° - 140°
16 - 20	16	16 (100%)	0 (0%)	75°	79° (21°)	45° - 120°
21 - 30	8	8 (100%)	0 (0%)	77.5°	81° (20°)	50° - 120°
> 30	5	5 (100%)	0 (0%)	90°	92° (18°)	70° - 120°
All examinations	408	399 (98%)	9 (2%)	60°	66° (29°)	5° - 140°

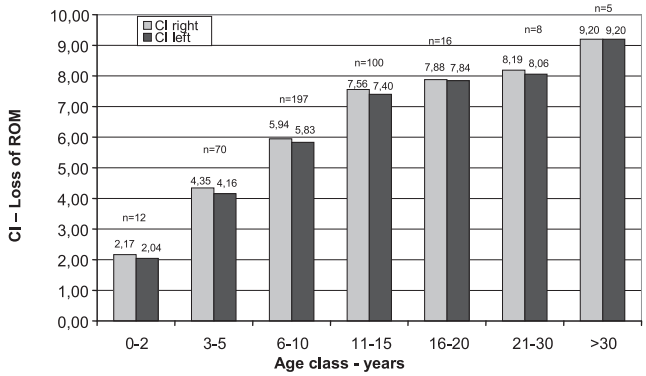


Fig. 2. Loss of range of motion (ROM) – relative contracture index (CI) – knee joint FL/EXT in SMA Type II

Tab. 3. Loss of range of motion (ROM) of ankle joint DF/PF in SMA Type II. Combined data for right and left limbs

Patients' age (years)	Number of examinations	Loss of ROM present (%)	No loss of ROM (%)	Loss of ROM [°]		
				Median	Mean (s_D)	Min - Max
0 - 2	12	7 (58%)	5 (42%)	50°	46° (14°)	20° - 65°
3 - 5	65	59 (91%)	6 (9%)	50°	53° (11°)	30° - 80°
6 - 10	185	177 (96%)	8 (4%)	55°	53° (12°)	10° - 80°
11 - 15	96	96 (100%)	0 (0%)	55°	56° (14°)	15° - 80°
16 - 20	12	12 (100%)	0 (0%)	52.5°	52° (16°)	25° - 75°
21 - 30	6	6 (100%)	0 (0%)	65°	63° (12°)	40° - 80°
> 30	5	5 (100%)	0 (0%)	70°	65° (9°)	50° - 75°
All examinations	381	362 (95%)	19 (5%)	55°	55° (13°)	10° - 80°

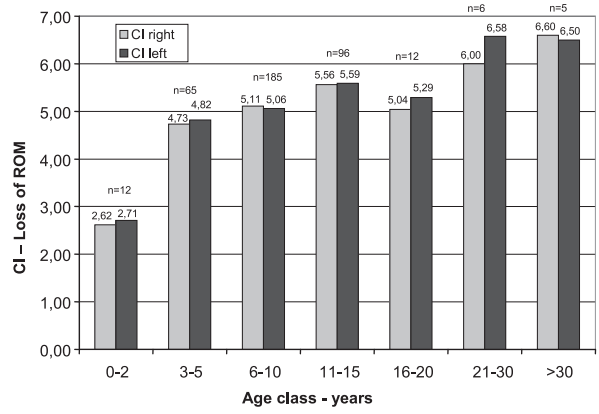


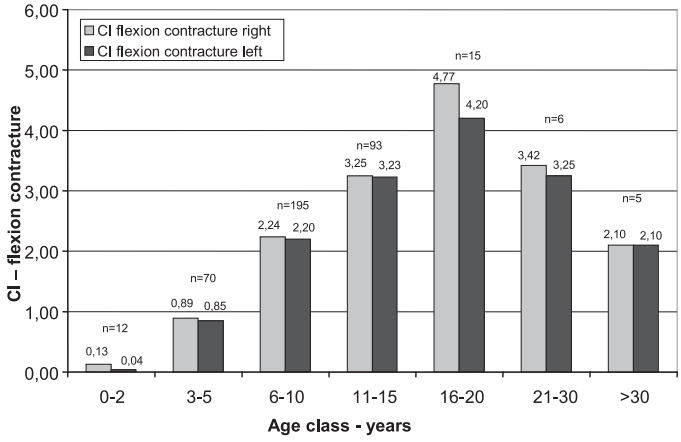
Fig. 3. Loss of range of motion (ROM) – relative contracture index (CI) – ankle joint DF/PF in SMA Type II

Tab. 4. Manifest flexion contracture of hip, knee and ankle joint in SMA Type II. Combined data for right and left limbs.

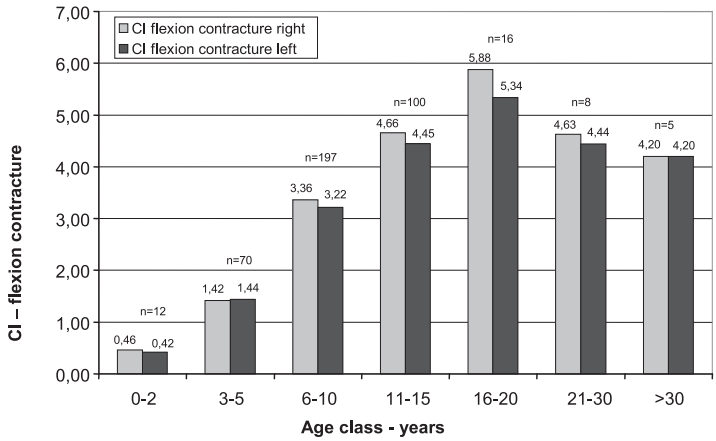
Patients' age (years)	Number of examinations	Flexion contracture present (%)	No flexion contracture (%)	Loss of ROM [°]		
				Median	Mean (s_D)	Min - Max
Hip joint						
0 - 2	12	1 (8%)	11 (92%)	10°	10° (7°)	5° - 15°
3 - 5	70	42 (60%)	28 (40%)	10°	15° (9°)	5° - 40°
6 - 10	195	165 (85%)	30 (15%)	25°	26° (14°)	5° - 80°
11 - 15	93	87 (94%)	6 (6%)	35°	35° (17°)	5° - 85
16 - 20	15	15 (100%)	0 (0%)	40°	45° (18°)	20° - 90°
21 - 30	6	6 (100%)	0 (0%)	37.5°	33° (13°)	10° - 50°
> 30	5	4 (80%)	1 (20%)	25°	26° (10°)	15° - 40°
All examinations	396	320 (81%)	76 (19%)	30°	27° (16°)	5° - 90°
Knee joint						
0 - 2	12	6 (50%)	6 (50%)	10°	10° (3°)	5° - 15°
3 - 5	70	61 (87%)	9 (13%)	10°	17° (13°)	5° - 70°
6 - 10	197	184 (93%)	13 (7%)	30°	36° (22°)	5° - 95°
11 - 15	100	98 (98%)	2 (2%)	40°	47° (23°)	5° - 90°
16 - 20	16	16 (100%)	0 (0%)	60°	56° (27°)	15° - 95°
21 - 30	8	8 (100%)	0 (0%)	40°	48° (23°)	10° - 90°
> 30	5	5 (100%)	0 (0%)	40°	42° (17°)	20° - 60°
All examinations	408	378 (93%)	30 (7%)	30°	37° (24°)	5° - 95°
Ankle joint (Equinus)						
0 - 2	12	2 (17%)	10 (83%)	10°	10° (4°)	5° - 15°
3 - 5	65	24 (37%)	41 (63%)	10°	14° (10°)	5° - 50°
6 - 10	185	50 (27%)	135 (73%)	10°	13° (9°)	5° - 40°
11 - 15	96	32 (33%)	64 (67%)	10°	13° (11°)	5° - 50°
16 - 20	12	4 (33%)	8 (67%)	5°	7° (3°)	5° - 10°
21 - 30	6	4 (67%)	2 (33%)	10°	19° (15°)	5° - 40°
> 30	5	4 (80%)	1 (20%)	10°	13° (8°)	5° - 20°
All examinations	381	120 (31%)	261 (69%)	10°	13° (10°)	5° - 50°

Tab. 5. Loss of overextension of hip and knee joint in SMA Type II. Combined data for right and left limbs

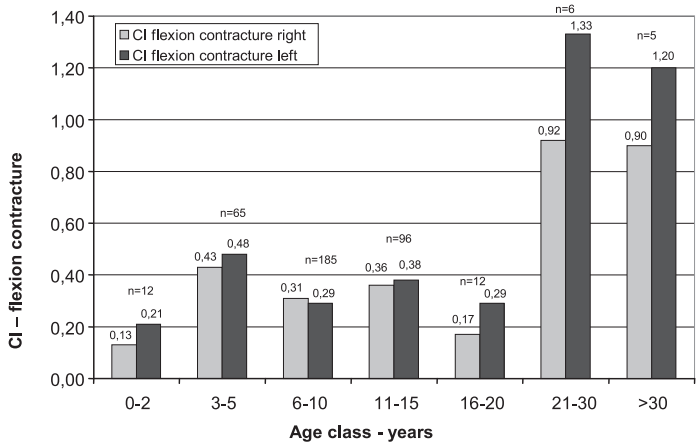
Patients' age (years)	Number of examinations	Loss of Over-EXT present (%)
Hip joint		
0 - 2	12	7 (58%)
3 - 5	70	56 (80%)
6 - 10	195	180 (92%)
11 - 15	93	91 (98%)
16 - 20	15	15 (100%)
21 - 30	6	6 (100%)
> 30	5	5 (100%)
All examinations	396	360 (91%)
Knee joint		
0 - 2	12	8 (67%)
3 - 5	70	66 (94%)
6 - 10	197	194 (98%)
11 - 15	100	100 (100%)
16 - 20	16	16 (100%)
21 - 30	8	8 (100%)
> 30	5	5 (100%)
All examinations	408	397 (97%)



Hip joint



Knee joint



Ankle joint

Fig. 4. Manifest flexion contracture of hip, knee and ankle joint – relative contracture index (CI) – in SMA Type II

13° (± 10°) (Table 4 and Diagram 4). The development of DF/PF clearly shows the increasing loss of range of motion, caused mainly by a decrease in plantar flexion.

DISCUSSION

This study shows that patients with SMA type II who are unable to walk frequently develop pronounced flexion contractures of the hip and knee joints as well as restricted mobility of the ankle joint with talipes equinus. The greatest limitations of extension (flexion contractures) appear in the hip. External and internal rotation appear only slightly restricted over the course of time. Greater restriction was observed in abduction/adduction, mainly caused by increased restriction of abduction with age.

The most pronounced contractures in the lower extremities appear in the knee joints already at a young age. Similar to the hip joint, physiological overextension of the knee had disappeared completely in over 60% of the patients by the age of 2.

Talipes equinus is caused by muscular weakness of the dorsal extensors. Restricted mobility of dorsal extension and talipes equinus contractures were also detected at an early age.

Our observations correlate with earlier studies [4 -8,10] but this examination of 143 patients represents the most extensive collection of data concerning joint contractures of the lower extremities in patients with SMA type II.

Johnson et al. examined 20 patients with chronic SMA at an average age of 37.9 years (1-75) and found restriction of hip extension (42% patients, mean 38°), knee extension (58% patients, mean 43°), and dorsal flexion of the ankle (47% patients, mean 29°). A shortening of the iliotibial band with restricted hip adduction of 30° was noted in one patient [6].

Wang et al. examined 27 patients with SMA type II from the age of 0.8 to 22.2 years (mean age 9.8± 6.5) [8]. They found the following restrictions: restriction of knee extension (89% patients, mean 48°), ankle dorsiflexion (52% patients, mean 25°), hip extension (48% patients, mean 40°), hip abduction (30% patients, mean 17°), hip flexion (15% patients, mean 18°), ankle plantar flexion (15% patients, mean 26°), hip internal rotation (2 patients, mean 13°), knee flexion (2 patients, mean 38°) and hip adduction (1 patient, 20°). Wang et al. also point out the very early onset of restriction of mobility of the joints of the lower extremities among infants.

Since the patient groups and methods in our study were different than in the previous studies, it is difficult to make a direct comparison of our CI values with those of Johnson et al. and Wang et al. but the highest CI values calculated across all studies (Johnson et al., Wang et al. and our study) were for the knee flexion contracture [6,8].

Benady collected the clinical findings of 50 children with intermediate and mild forms of SMA (SMA type II and III). There were mostly young children in this study, their ages at the last examination ranging between 13 months and 18 years. Benady describes ankle joint contractures with equinus deformity in 45% (21 of 47) of the children. Seven and five children respectively presented with knee and hip flexion contractures [5].

Carter et al. examined joint mobility in 18 patients with SMA type II at a mean age of 17 years (±14). They found restriction of mobility of at least 20° in the knee joint of 50% of the patients, in the hip joint of 38% and in the ankle of 22% of the patients [7].

Evans et al. examined 21 patients with SMA II for an average of 15 years after their condition was diagnosed at the average age of 7 months. They observed an increase of hip and knee flexion contractures of up to 80° at the age of 10. The equinus deformity was noted in about half their patients [4].

Bono et al. examined 20 children with SMA II aged between 3.1 and 15.7 years. Among other things, they investigated the prevalence of lower limb contractures. They observed mild contractures (< 20°) at the age of 5 in a total of 10 patients. At the age of 10, 70% of the children had mild contractures and 30% had severe contractures (> 20°) [10].

The patient populations described in other published papers are much smaller in number than ours. Their data were collected and presented quite differently and are to an extent relatively incomplete in comparison to our study.

Our study does contain some methodological weaknesses. The combination of longitudinal and cross-sectional data means that we are limited in our prediction of the actual individual potential for the development of contractures.

This study includes more than one evaluation from each patient and a few patients can dominate the take-home line.

If one patient was evaluated several times during a particular age period and he manifested the abnormality only at the last evaluation, this could influence the data as the probability of recording an abnormal finding in that patient was higher compared to the individual who was seen only once at the beginning of the period. The choice for our evaluation of data from examinations with the greatest loss of range of motion could lead to subject selection, which would tend to magnify the effect of error and tend to bias the result toward greater impairment.

Because we divided the patients into age groups and because of the number of examinations, we were able to make more precise statements about those aged about 3 to 15 years. Younger and older patients had been examined fewer times so we could estimate only roughly how the contractures would develop.

Despite these methodological problems, the insight we have gained into the development of contractures of the lower extremities in SMA II represents an important scientific contribution to understanding the natural course of this condition.

Of particular value in this case is the unusually big group of patients and the large data collection from the examinations.

To date there has been no scientific information about conservative (e.g. physiotherapy, orthoses) vs. surgical treatment of contractures of the lower extremities in patients with SMA type II. Further studies of this subject are needed.

CONCLUSIONS

1. This study shows the development of restrictions of passive range of motion in the joints of the lower extremities across the age groups in SMA type II patients.
2. Loss of range of motion and contractures of the joints of the lower extremities (hip, knee and ankle) were seen in many patients at a very early age (in many cases under the age of 2 years) and progressively increased with age in childhood.
3. The greatest restriction of motion with flexion contractures was in the knee joint followed by hip and ankle.
4. The results of this study may be used as a basis for comparisons in further studies dealing with the effectiveness of treatments to address both the contracture itself as well as the underlying cause of the contracture in SMA.

REFERENCES

1. Zerres K, Rudnik-Schöneborn S, Forrest E, Lusakowska A, Borkowska J, Hausmanowa-Petrusewicz I. A collaborative study on the natural history of childhood and juvenile onset proximal spinal muscular atrophy (type II and III SMA): 569 patients. J Neurol Sci 1997; 146: 67-72.
2. Forst R, Ingenhorst A, Mortier W. Neuromuskuläre Systemerkrankungen. In: Zichner L editor. Orthopädie und Orthopädische Chirurgie. Systemerkrankungen. Stuttgart New York: Thieme; 2003. 243-254.
3. Fujak A, Wollinsky KH, Forst R. Proximal spinal muscular atrophy (SMA). Z Orthop Unfall 2007; 145: 233-252.
4. Evans GA, Drennan JC, Russman BS. Functional Classification and orthopaedic management of spinal muscular atrophy. J Bone Joint Surg [Br] 1981; 63: 516-522.
5. Benady SG. Spinal muscular atrophy in childhood: review of 50 cases. Dev Med Child Neurol 1978; 20: 746-757.
6. Johnson ER, Fowler WM Jr, Liebermann JS. Contractures in neuromuscular disease. Arch Phys Med Rehabil 1992; 73: 807-810.
7. Carter GT, Abresch RT, Fowler WM Jr, Johnson ER, Kilmer DD, McDonald CM. Profiles of neuromuscular diseases. Spinal muscular atrophy. Am J Phys Med Rehabil 1995; 74: 150-159.
8. Wang HY, Ju YH, Chen SM, Lo SK, Jong YJ. Joint range of motion limitations in children and young adults with spinal muscular atrophy. Arch Phys Med Rehabil 2004; 85: 1689-1693.
9. Breusch S, Mau H, Sabo D. Klinikleitfaden Orthopädie. 4th Edition. München Jena: Urban & Fischer; 2002.
10. Bono R, Inverno M, Botteon G, Iotti E, Estienne M, Berardinelli A, Lanzi G, Fedrizzi E. Prospective study of gross motor development in children with SMA type II. Ital J Neurol Sci 1995;16 : 223-230.

Liczba słów/Word count: 3886 **Tabele/Tables:** 5 **Ryciny/Figures:** 4 **Piśmiennictwo/References:** 10

Adres do korespondencji / Address for correspondence
Dr. med. Albert Fujak, Department of Orthopaedic Surgery, Friedrich-Alexander-University Erlangen-Nuremberg, Rathsberger Str. 57, D-91054 Erlangen, Germany,
Tel.: + 49 9131 822 3303Fax: + 49 9131 852 3565, e-mail: A.Fujak@t-online.de

Otrzymano / Received 10.05.2010 r.
Zaakceptowano / Accepted 15.09.2010 r.