

Scoliosis in Rett syndrome – natural history and surgical treatment

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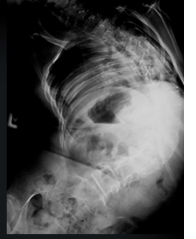


ICEOS

5th International Congress on
Early Onset Scoliosis
and Growing Spine

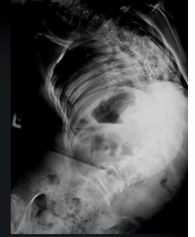
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Introduction



- › **Rett syndrome (RS)** is a rare genetic disorder affecting only girls
 - › Mutation of MCEP 2 gene located in chromosome X¹
 - › Prevalence is 1:15000¹
 - › Diagnosis based on characteristic clinical findings and genetic tests²
 - › Normal development occurs until 12-18 months, following that a regression in physical and mental status is typical
 - › **Orthopedic aspects:** scoliosis, contractures, foot deformities, hip luxation

Introduction - scoliosis

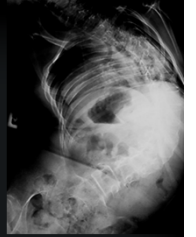


- › Prevalence of scoliosis in patients with RS varies from 36%-100% of patients depending on the neurological status³⁻⁵
- › Typically scoliosis is diagnosed at the age 8 – 10 years, progression may occur despite skeletal maturity^{4,5}
- › Annual curve progression may reach up to 14° -21°⁶

Aim of paper

- › Describe the preoperative curve behavior in a series of patients
- › Analyze course of surgery
- › Analyze radiologic parameters
- › Assess subjectively the results

Material

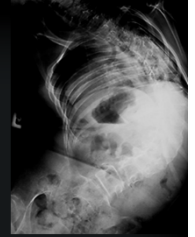


- › 13 girls with RS diagnosis
- › 10 with scoliosis
- › 9 girls treated surgically – study group
- › Age at surgery 11.7 years (9-16 years)
- › Follow-up 3.1 years (1-6 years)

Methods

- › Radiological data (curve type, apical vertebral translation, coronal and sagittal balance) measured pre- and postoperatively
- › Surgical data (type of surgery, course of surgery and early postoperative period)
- 4 › Subjective evaluation (questionnaire handled to caregivers)

Results

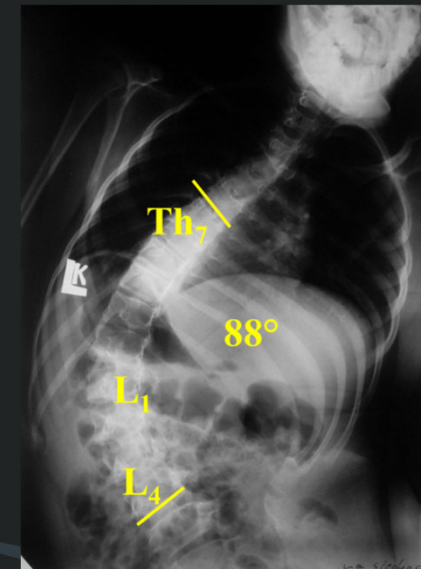


Basic data:

- Age of scoliosis onset **9.3 years** (5-13 years)
- 8 girls non-ambulant, 1 walking with aid
- 7 treated with rigid brace, only 2 (22.2%) compliant
- Preoperative follow-up **12.5 months** (6 – 24 months)

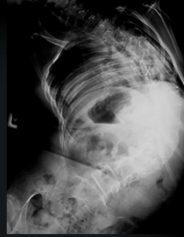
Curve types:

Nr	Lenke type	Curve apex	Nr of vertebrae in the curve	Ambulant (A)/ Non-ambulant(NA)
1	5C+	T12	11	NA
2	5CN	T12	7	NA
3	1A+	T9	9	A
4	5C+	T12	10	NA
5	5C+	L2	5	NA
6	5C+	L1	10	NA
7	5C+	T12	11	NA
8	1A+	T11	9	A
9	5C+	T12	10	NA



Girl, S.N., age 11, 5CN+ curve type

Results



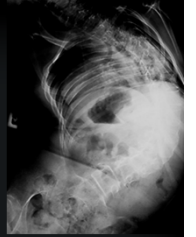
› Preoperative change in the radiological parameters:

	Cobb angle change (°)	C7-CSVL change (mm)	Pelvis inclination change (°)	Thoracic kyphosis change (°)
Mean change	16.1°	15.3 mm	7.1°	12.6°
Range	5-35°	4-45mm	-2-15°	-5-35°

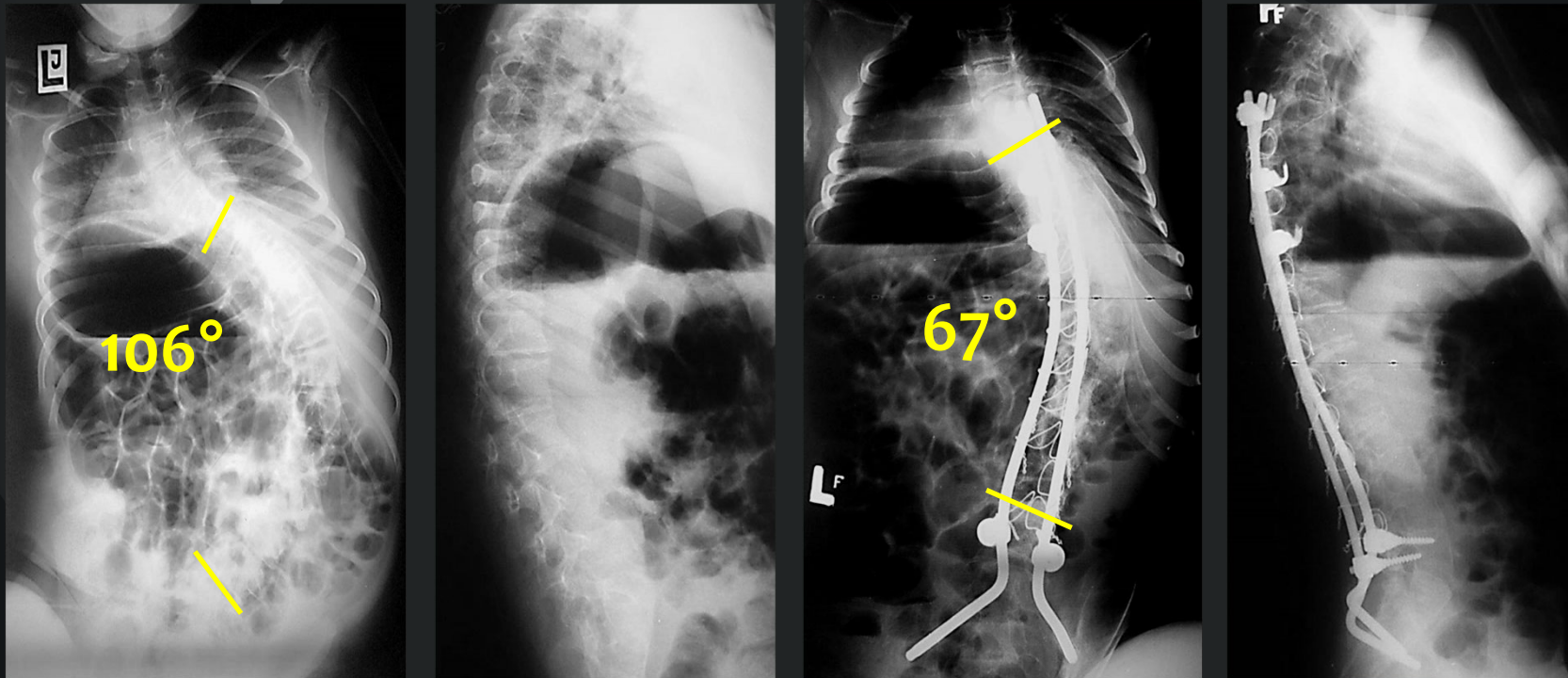
› Surgical procedure:

- › 6 girls – posterior fusion Galveston technique
- › 1 girl – posterior selective fusion, pelvis not included
- › 2 girls – anterior fusion (1 ambulant)
- › Time of surgery – 126 minutes (100-250)
- › Blood loss – posterior fusion 650 ml, anterior fusion 250 ml
- › Prolonged mechanical ventilation – 5 girls (24 hours)

Results

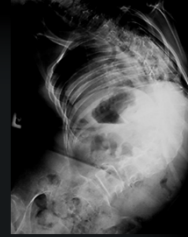


› Surgical procedure



**Patient P.M., age at surgery 12 years. Non-ambulant.
Posterior fusion Galveston technique, with pedicular screws and hooks.
Good result.**

Results

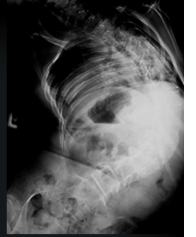


› Radiological results:

	Cobb angle pre-op(°)	Cobb angle post-op(°)	Correction post-op (%)	Correction follow-up(%)
Mean change	85°	54.1°	38%	33%
Range	52-120°	18-85°	20-67%	11-58%

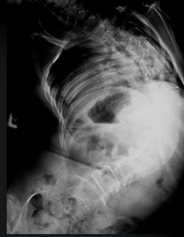
	Pre-op	Post-op	Follow-up	Correction follow-up(%)
AVT	63.3mm	25.6mm	26.7mm	53.8%
C7-CSVL	43.9mm	33.3mm	25.6mm	25.5%
Pelvic inclination	30°	10.7°	13°	62.4%
T4-T12 kyphosis	69.7°	45°	44°	33.8%
T10-L2 junction	25.9°	22.4°	21.2°	5.4%

Results



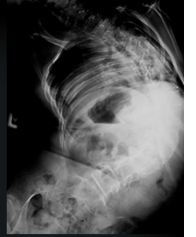
- › **Subjective results:**
- › 6 caregivers (66.7%) agreed to participate in the questionnaire
- › All girls non-ambulant, operated with the same technique
- › **Cosmetic effect** 5/6 (83.3%) – very good,
1/6 (16.7%) – not changed
- › **Function** 3/6 (50%) – better,
2/6 (33%) – not changed,
1/6 (16.7%) – worse than preoperatively
- › **Recommend surgery** 6/6 (100%) – yes
- › **Additional remarks** 4/6 (66.7%) – hesitated too long to undergo surgery

Conclusions



- › **Curves are usually long-sweeping, but in ambulant patients may resemble idiopathic-like curves**
- › **Rate of scoliosis progression in patients with Rett syndrome is high**
 - › **The initial curve magnitude is a risk factor of curve progression**
- › **Deformity may be effectively treated surgically**
 - › **Type of surgery depends on curve type and neurological status**
- › **There are no major risks of surgery**
- › **The results of surgery during follow-up period are stable**

References



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