Evaluation of Syndromic Spinal Deformities

Mehmet B.Balioglu[1], MD; Mehmet T.Tacal[2], MD; Akif Albayrak[2], MD, Secil S.Sakizlioglu[3], MD

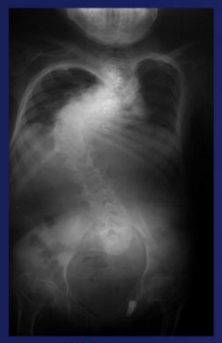
[1]Departman of Orthopaedic Surgery, Medical Faculty of Kafkas University, Kars

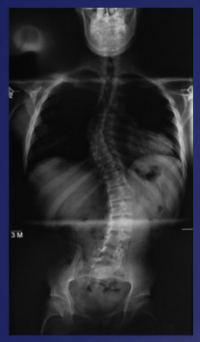
[2] Departman of Orthopaedic Surgery, MS Baltalimani Bone Disease Teaching Hospital, Istanbul

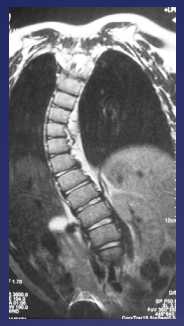
[3] Departman of Radiology, MS Baltalimani Bone Disease Teaching Hospital, Istanbul

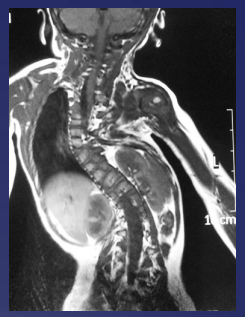
Introduction

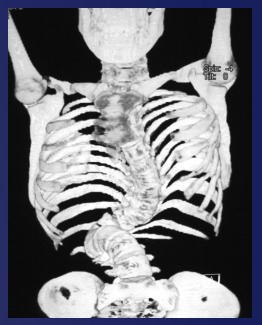
- Patients with the rare scoliosis condition of syndromic spinal deformity were evaluated to determine both spine related problems and other anomalies.
- Clinical and radiological examinations of patients were reviewed to assess related anomalies.


KK-Larsen


SED

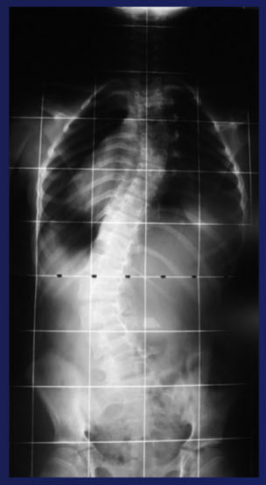

AT-Escobar

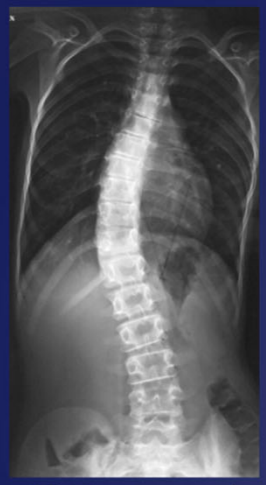

IDN-Jarcho-Levin Syndrome

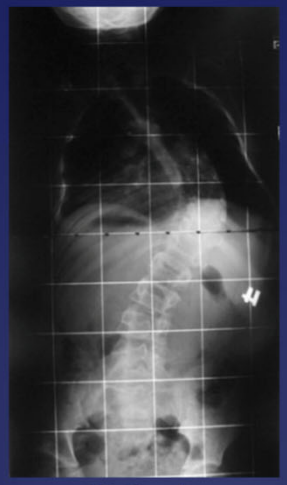

NK-Marfan

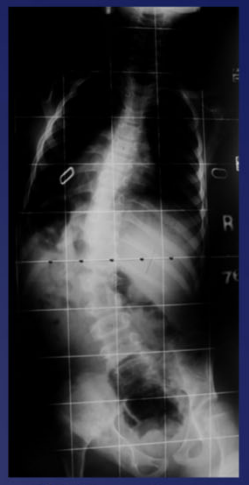
Method

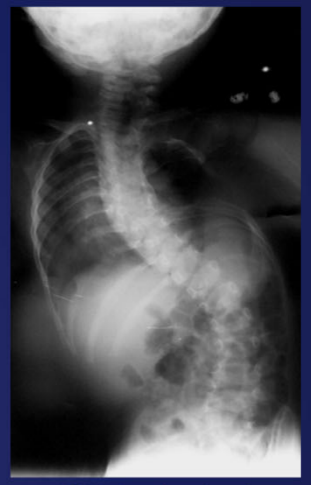
- 22 patients were diagnosed with syndromic scoliosis (SS) by genetic analysis.
- The average age was 13.2 (3.2-19.6) 13 male, 9 female.


BK-9 P Syndrome


EO-Eisenmenger's


AT-Escobar


SG-Genoderma


IDN-Jarcho-Levin

Method

- All patients received clinical examinations, including full neurological and functional evaluation of all extremities, and when appropriate magnetic resonanance imaging (MRI) and computed tomography and genetic analysis.
- Cardiovascular and urinary systems were also evaluated.
- For some patients endocrinologic and neurosurgical examinations were conducted.

Result

- Marfan 3,
- Neurofibromatosis 2 patients,
- Escobar 2,
- HSMN type IV 2,
- Genoderma Osteodistfrica 2,
- Spondyloepiphysial dysplasia 1,
- Larsen 1,
- Digorge syndrome 1,
- Eisenmenger 1,
- Gilbert 1,
- 9p 1,
- 1p+ 1,
- Marquio 1,
- Rubinstein Taybi 1,
- Desbuguois 1,
- Pierre Robin Sekans 1.

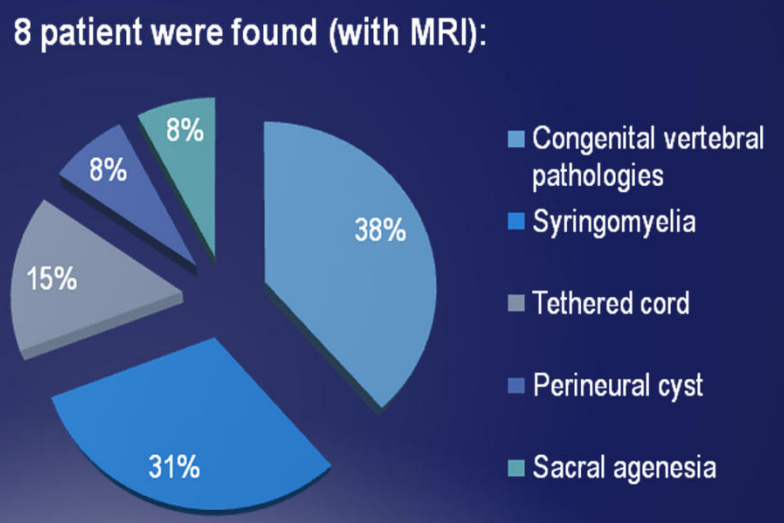
Result

- Cobb angles revealed the scoliotic curve average: 33° (11°-83°), and kyphotic with a maximum of 84°.
- Only 15 patients were evaluated with MRI.
- Seven were found normal.

Syndrome	Number	Age (y)	Gender	Cobb Angle
Neurofibromatosis	2	13.2 (3.2-19.6)	13 Male 9 Female	33° (11-83)
Marfan	3			
Escobar	2			
HSMN type IV	2			
Spondyloepiphysial dysplasia	1			
Genoderma Osteodistfrica	2			
Larsen, Digorge, Eisenmenger, Gilbert, 9p, 1p+, Marquio, Rubinstein Taybi, Desbuguois, Pierre Robin Sekans	1			

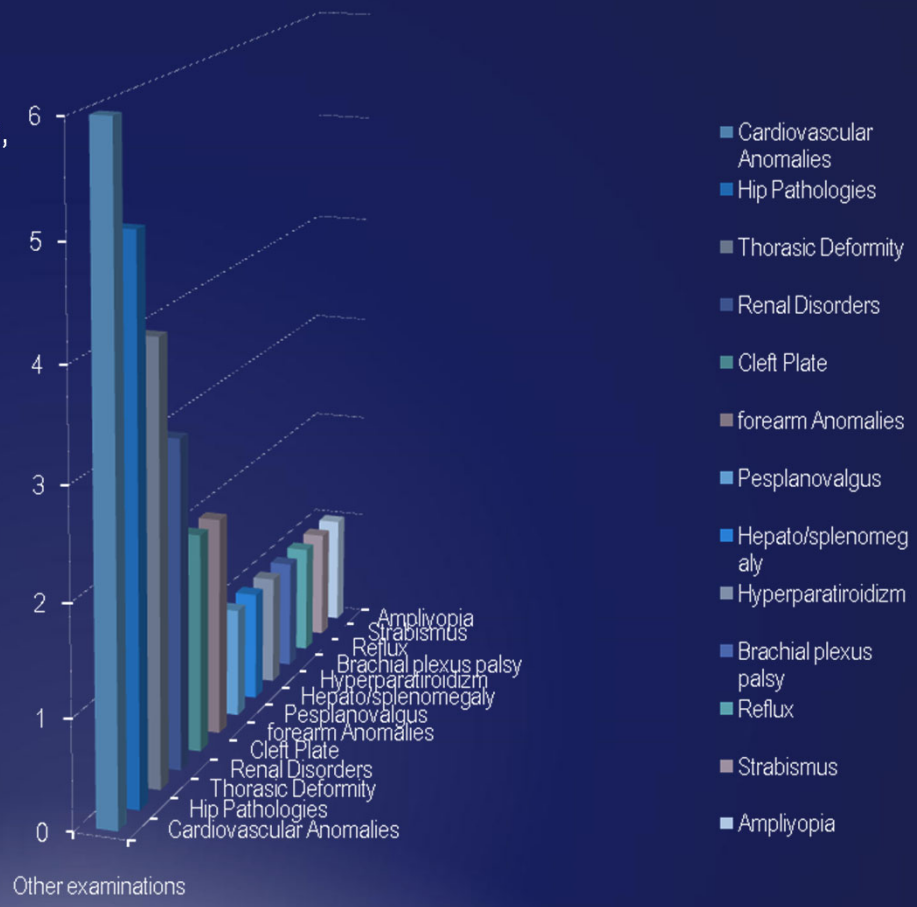
Result

- Of the remaining 8 patients:
  - Congenital vertebral pathology (5): the most common pathology,
  - Syringomyelia (4),
  - Tethered cord (2),
  - Perineural cyst (1),
  - Sacral agenesia (1).



Result

- Other examinations revealed
  - 6 patients had cardiovascular anomalies,
  - 5 hip pathologies,
  - 4 thoracic deformity,
  - 3 renal disorders,
  - 2 cleft palate,
  - 2 forearm pathologies,
  - 2 pesplanovalgus,
  - 1 hepatomegaly/splenomegaly,
  - 1 hyperparathyroidism,
  - 1 brachial plexus palsy,
  - 1 reflux,
  - 1 strabismus,
  - 1 amblyopia.



Conclusion

- MRI analysis revealed that syringomyelia and congenital vertebral anomalies were the most common pathologies in patients with SS.
- The majority of concomitant pathologies in these patients were cardiovascular, nephrological and hip disorders.