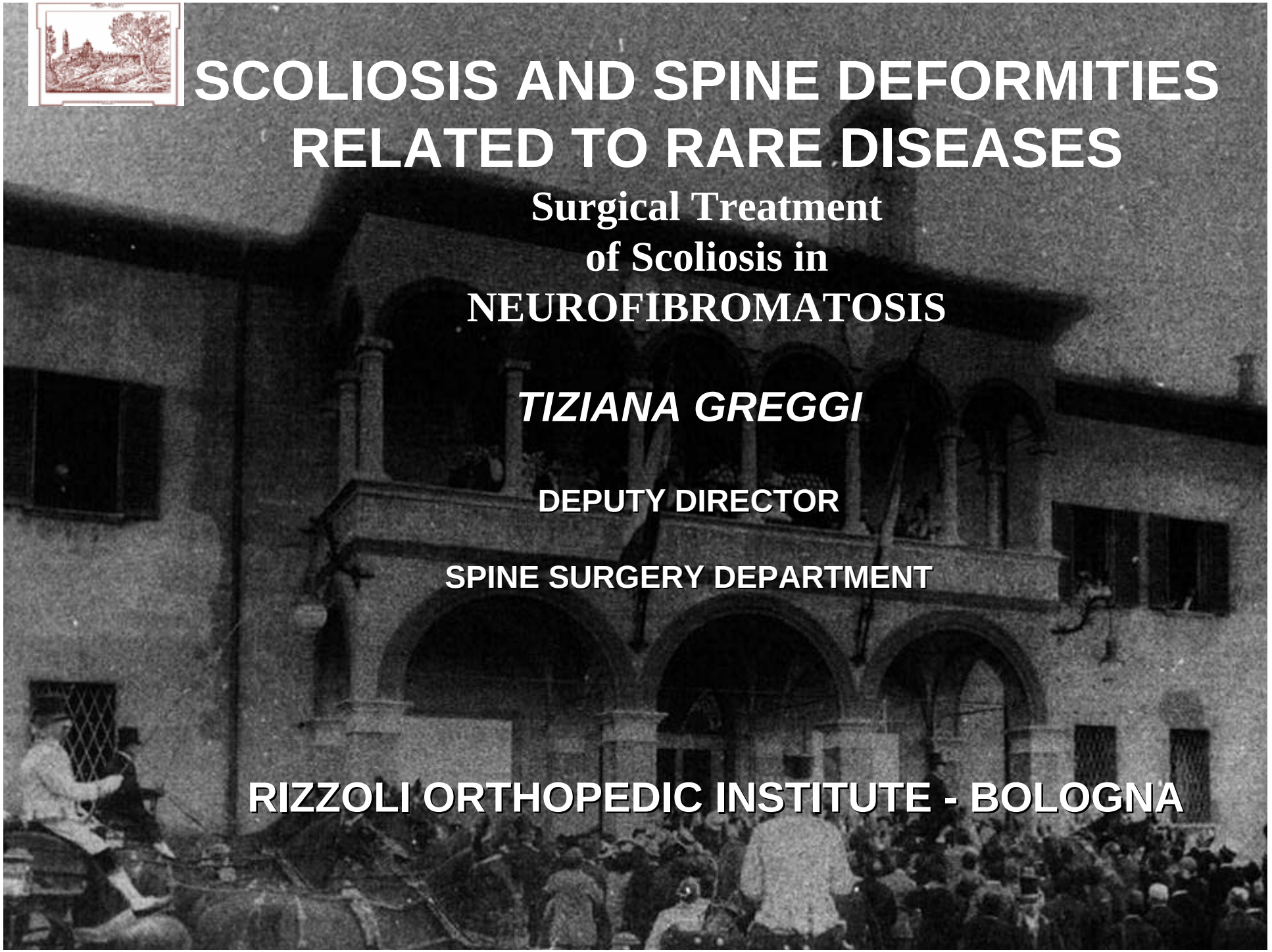


SCOLIOSIS AND SPINE DEFORMITIES RELATED TO RARE DISEASES

Surgical Treatment of Scoliosis in NEUROFIBROMATOSIS

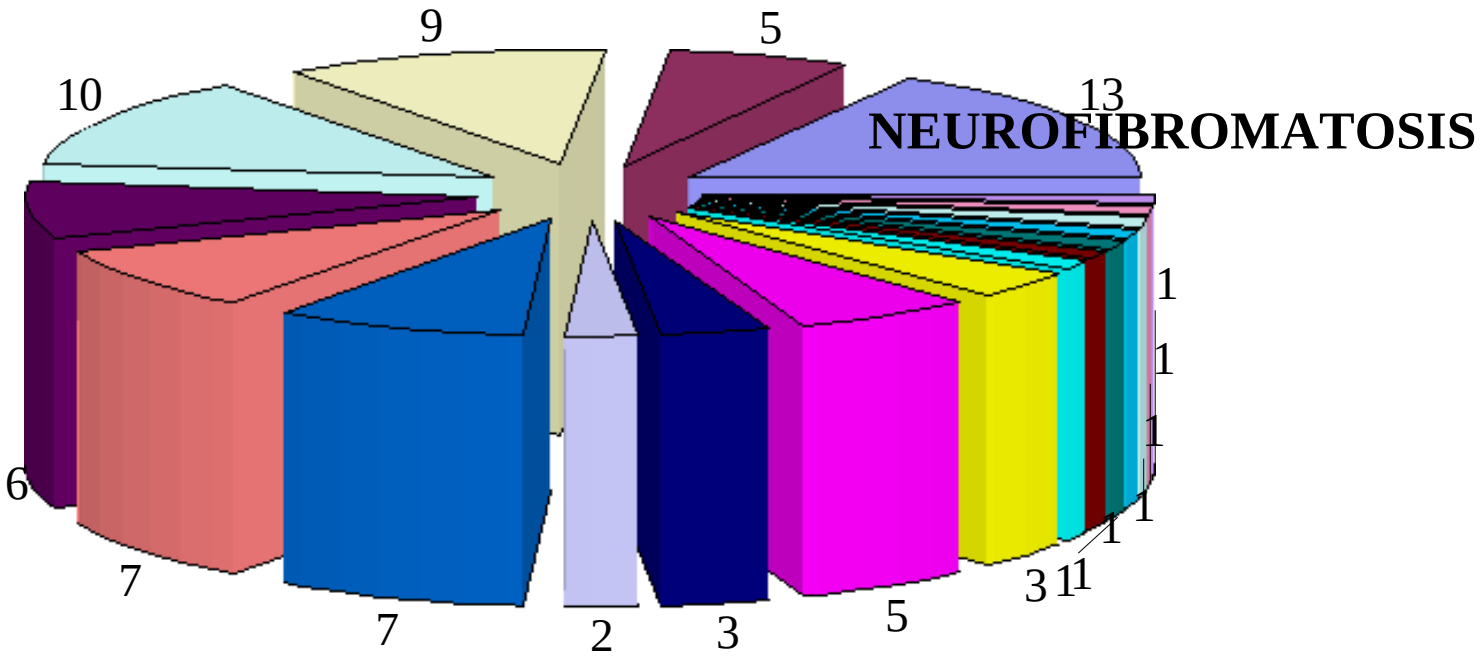
TIZIANA GREGGI

DEPUTY DIRECTOR

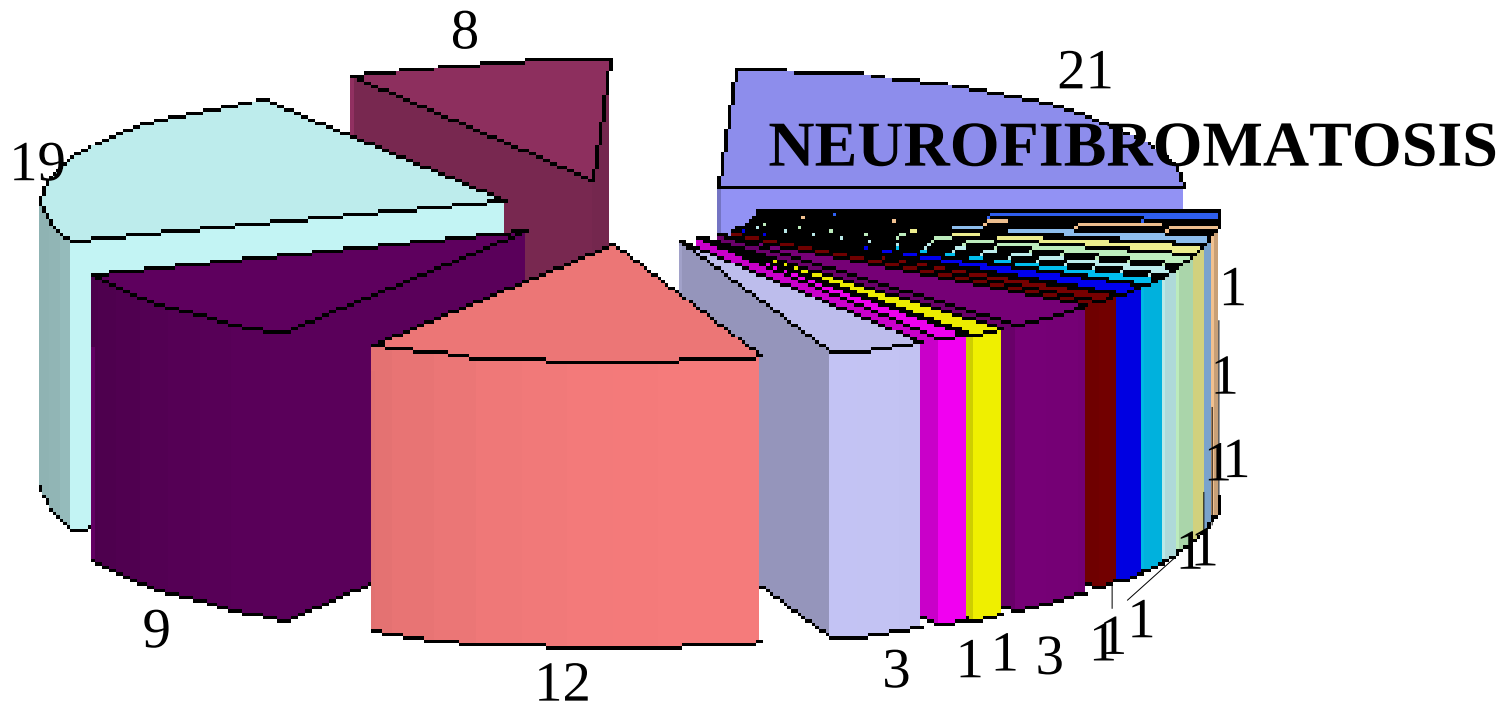
SPINE SURGERY DEPARTMENT

RIZZOLI ORTHOPEDIC INSTITUTE - BOLOGNA

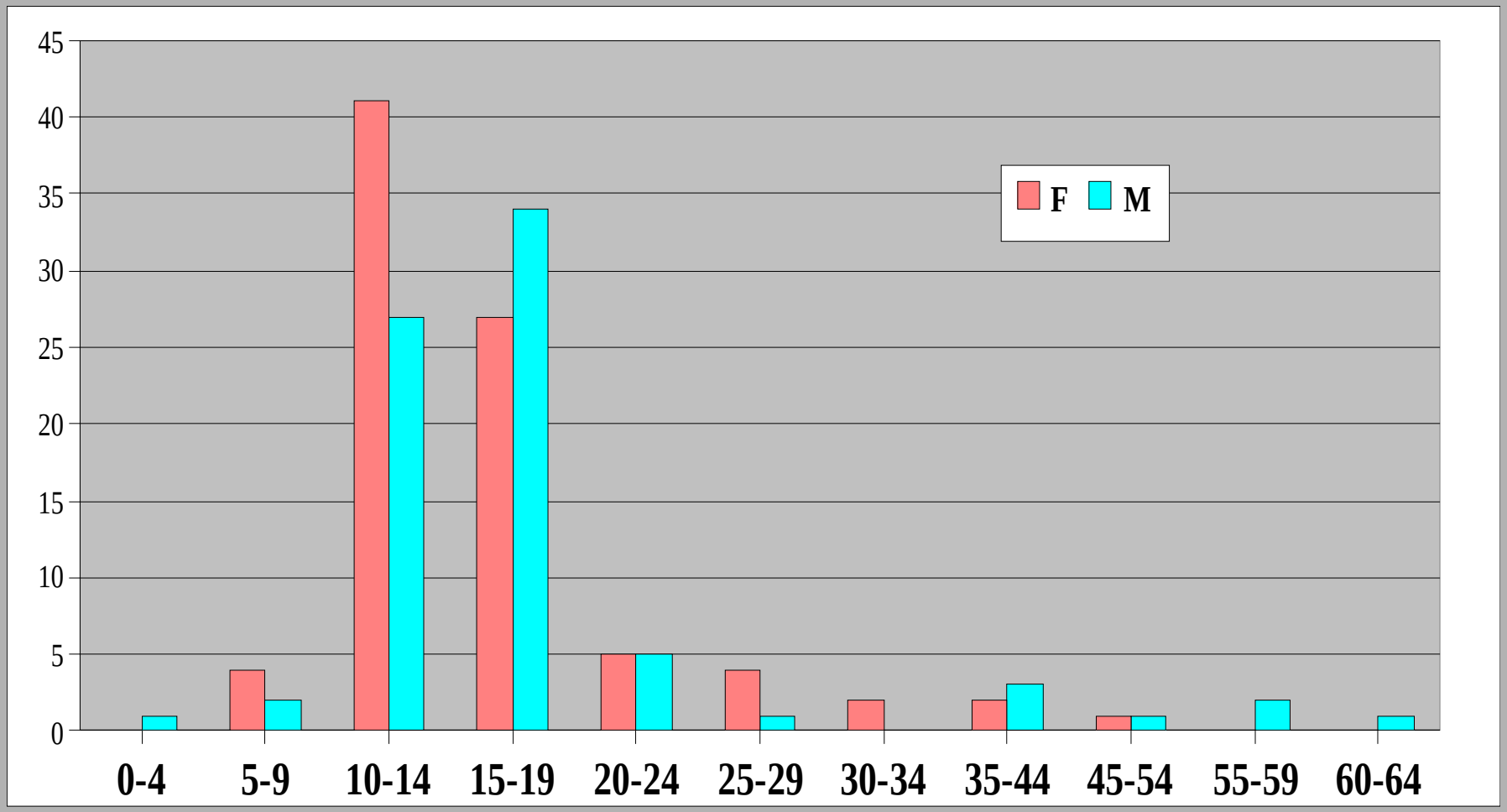
RARE DISEASES IN MALES



RARE DISEASES IN FEMALES



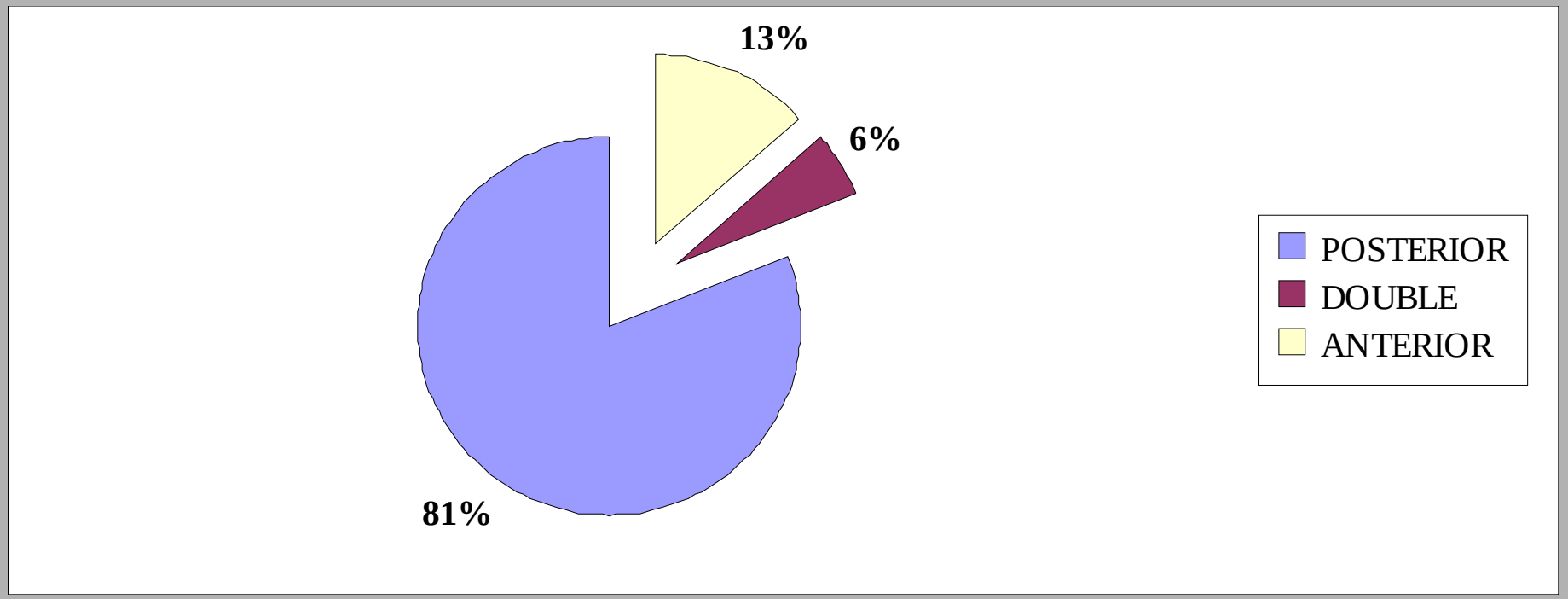
PATIENTS BY SEX AND AGE AT FIRST SURGICAL INTERVENTION



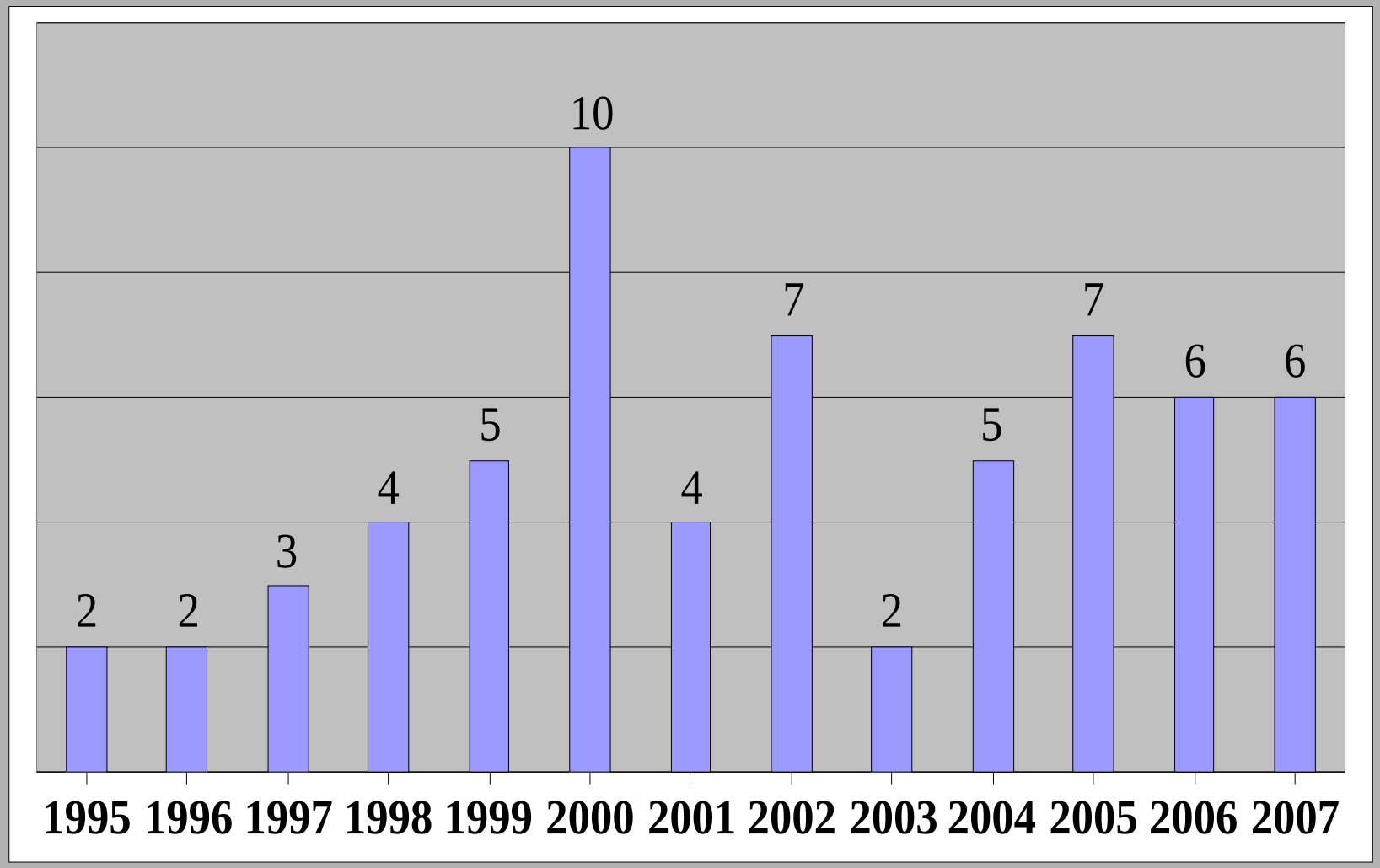
NUMBER OF PATIENTS BY REGION OR FOREIGN COUNTRY OF ORIGIN (2006)

	SEX		
	F	M	Total
CAMPANIA	9.88%	13.24%	11.41%
EMILIA ROMAGNA	9.88%	13.24%	11.41%
LOMBARDY	7.41%	13.24%	10.07%
PUGLIA	7.41%	11.76%	9.40%
TUSCANY	9.88%	4.41%	7.38%
SARDINIA	3.70%	8.82%	6.04%
LAZIO	6.17%	4.41%	5.37%
SICILY	7.41%	2.94%	5.37%
CALABRIA	6.17%	2.94%	4.70%
VENETO	3.70%	5.88%	4.70%
FRIULI V.G.	3.70%	4.41%	4.03%
PIEDMONT	4.94%	2.94%	4.03%
UMBRIA	6.17%	0.00%	3.36%
BASILICATA	1.23%	2.94%	2.01%
TRENTINO	1.23%	2.94%	2.01%
ABRUZZO	1.23%	1.47%	1.34%
SERBIA-MONTENEGRO	1.23%	1.47%	1.34%
ALBANIA	1.23%	0.00%	0.67%
INDIA	1.23%	0.00%	0.67%
LIBIA	0.00%	1.47%	0.67%
LIGURIA	0.00%	1.47%	0.67%
MOLDAVIA	1.23%	0.00%	0.67%
ROMANIA	1.23%	0.00%	0.67%
TUNISIA	1.23%	0.00%	0.67%
UKRAINE	1.23%	0.00%	0.67%
MARCHE	1.23%	0.00%	0.67%
Total	100.00%	100.00%	100.00%

KINDS OF SURGICAL ACCESS



NUMBER OF INTERVENTIONS AT THE FIRST TRIMESTER OF EACH YEAR




Foreign countries:
6.71%
Valle
0%
Trentino
2.01%
Friu
4.03%
Lombardia
10,07%
Veneto
4,70%
Piemonte
4.03%
Er
11.41%
magna
0.67%
Liguria
7.38%
Toscana
Marche
0.67%
3.36%
Umbria
Abruzzo
1,34%
Molise
0%
Lazio
5.37%
Puglia
9.40%
Campania
11,41%
2.01%
6.04%
Sardegna
Basilicata
Calabria
4.70%
Sicilia
5.37%

Von Recklinghausen's disease

Autosomal dominantgenetic disorder

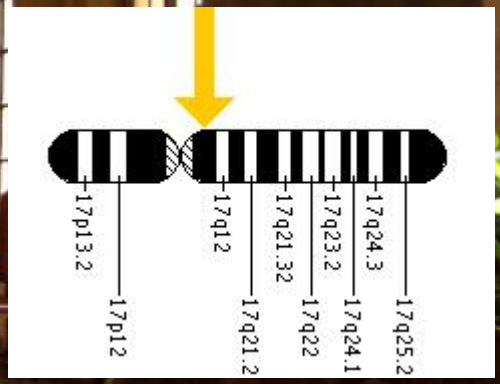
Where is the NF1 gene located?

Cytogenetic Location: 17q11.2

Molecular Location on chromosome 17: base pairs 26,446,242 to 26,725,589

1/5000 individuals

It is inherited as an autosomal dominant trait, but about 50 percent of cases arise as mutations.



EXCELLENCE MEDICINE
NOT
EVIDENCE BASE MEDICINE

Parisini P, Di Silvestre M, Greggi T, Paderni S, Cervellati S, Savini R.
Surgical correction of dystrophic spinal curves in neurofibromatosis. A review of 56 patients.
Spine. 1999 Nov 1;24(21):2247-53.

Li M, Fang X, Li Y, Ni J, Gu S, Zhu X.
Successful use of posterior instrumented spinal fusion alonefor scoliosisin 19 patients with neurofibromatosis type-1 followed up for at least 25 months.
Arch Orthop Trauma Surg. 2008 Jul 24.

Study Design. A presentation of the results from 56 patients with dystrophic spinal deformities caused by neurofibromatosis surgicallymanaged from1971 to 1992.
Objectives. To focus on the need for combined anterior and posterior fusion in the presence of severe spinal dystrophic changes.
Summary of Background Data. It has been stated that the most effective management for dystrophic curves is early and aggressive surgery.
Methods. The patients were divided into two groups: Type I scoliosis (kyphosis ,50°) and Type II kyphoscoliosis (kyphosis .50°). Results were evaluated in relation to the type of surgery performed: single posterior instrumented fusion or preplanned combined anterior and posterior fusion.
Results. At a mean follow-up period of 15 years (range, 5–22 years), all patients appeared to be stabilized, after a total of 120 surgical interventions. In Group I, the posterior instrumented fusion failed in nine patients (47%), and in Group II it failed in seven patients (63%). The
preplanned combined anterior and posterior fusion failed in two patients (33%) in Group I and in four patients (20% in Group II. The failure incidence of the posterior instrumented fusion alone and of the planned anterior and posterior fusion was 53% (16 patients) and 23% (6 patients), respectively.
Conclusions. The severe dystrophic curve with anterior vertebral scalloping always requires combined anterior and posterior stabilization, particularly in younger patients, even if the sagittal curves have not become pathologic by the time of presentation. [Key words: neurofibromatosis, spine dystrophic deformity, spine fusion] Spine 1999;24:2247–2253

56
patients with dystrophic spinal deformities caused by
neurofibromatosis surgically managed from1971 to 1992.

NUMBER OF PATIENTS:
FROM 1993 TO 2007 ➢ 34

NEUROFIBROMATOSIS 100 CASES

NEUROFIBROMATOSIS (FROM 1993 TO 2007)

NUMBER OF PATIENTS BY SEX:

- **M: 13**
- **F: 21**

MEAN AGE: 16.9 yrs. (dev. st.: 9.4 yrs.)

- **M: 17.1 (dev. st.: 8.2)**
- **F: 16.9 (dev. st.: 10.0)**

NUMBER OF PATIENTS SUFFERING FROM NEUROFIBROMATOSIS BY REGION OR FOREIGN COUNTRY OF ORIGIN (2007)

	F	M	Total
CAMPANIA	9.52%	15.38%	11.76%
EMILIA ROMAGNA	14.29%	7.69%	11.76%
LOMBARDY	9.52%	7.69%	8.82%
PUGLIA	4.76%	15.38%	8.82%
TUSCANY	9.52%	7.69%	8.82%
SARDINIA	0.00%	15.38%	5.88%
LAZIO	9.52%	0.00%	5.88%
SICILY	4.76%	0.00%	2.94%
CALABRIA	4.76%	0.00%	2.94%
VENETO	0.00%	23.08%	8.82%
PIEDMONT	9.52%	7.69%	8.82%
UMBRIA	4.76%	0.00%	2.94%
ABRUZZO	4.76%	0.00%	2.94%
ALBANIA	4.76%	0.00%	2.94%
UKRAINE	4.76%	0.00%	2.94%
MARCHE	4.76%	0.00%	2.94%
Total	100.00%	100.00%	100.00%

NEUROFIBROMATOSIS (FROM 1993 TO 2007)

KINDS OF SURGICAL ACCESS:

- 8 DOUBLE

KINDS OF SURGICAL ACCESS:

- 39 POSTERIOR

KINDS OF SURGICAL ACCESS:

- 15 ANTERIOR

Growing SPINE

1. Not FUSION

(Johnston et al. 2008, Vitale et al. 2008)

2. Correction withougt fusion

- "Growth Sparing" (< 8 ANNI)
  - -Rib based (VEPTR) (Campbell et al)
  - -Spine based (DUAL ROD) (Akbarnia et al)

- "Growth Modulating" (> 8 ANNI)
  - -Staples (Betz et al)
  - -Shilla technique (McCarthy et al)
  - -Spinal tethering (Newton et al)

David Skaggs, M.D., Personal Comunication: 2nd International Congress on Early Onset Scoliosis & Growing Spine (ICEOS) Montreal, Canada, November 7-8, 2008

Surgical Treatment of Scoliosis in Neurofibromatosis

Rizzoli Ortopedic Institute

Bologna

Vertebral Surgery Division

Neurofibromatosis type 1 (NF-1) is a multisystemic disease. It may manifest as abnormalities of the nervous tissue, bones, soft tissue, and skin. The manifestations of NF-1 vary from person to person and range from subclinical to severe. Individuals who carry the gene eventually exhibit some clinical feature of the disease. The penetrance for NF-1 nears 100% during adulthood. Skeletal abnormalities are common in NF-1, with most patients presenting with some type of bony dysplasia. The orthopedic complications usually appear early. They include spinal deformities, such as scoliosis or kyphosis, congenital tibial dysplasia with bowing and pseudarthrosis of the tibia, forearm, other bones, as well as overgrowth phenomenon of an extremity, and soft tissue tumors.

Surgical Treatment of Scoliosis in Neurofibromatosis

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2 subtypes: idiopathic and dystrophic;

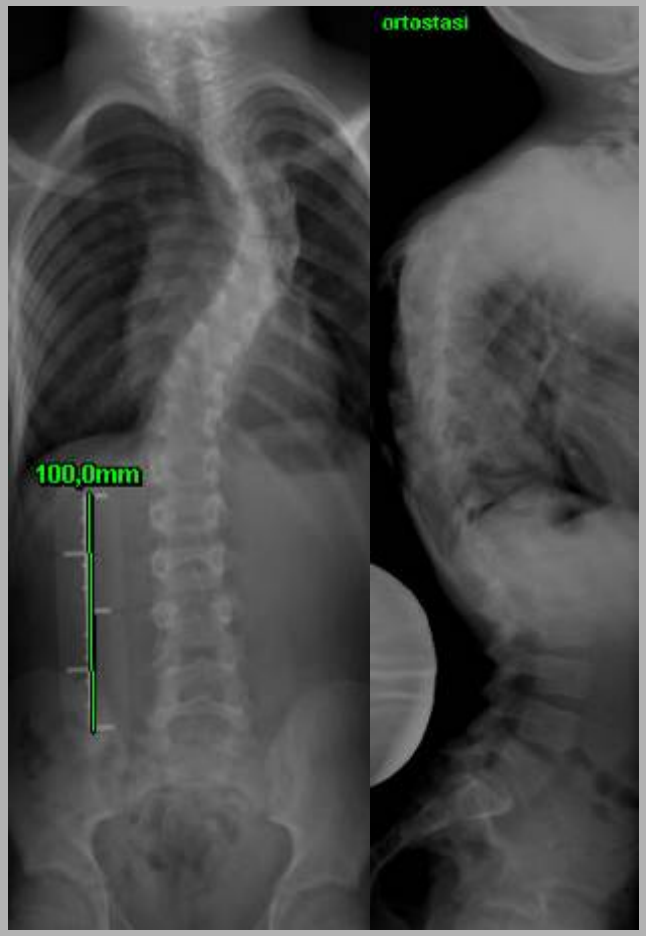
- dysplastic type: (dystrophic) Winter 80%
  - short, sharply angulated curve which involve only few vertebra;
  - associated w/:
    - neural foramina enlargement
    - rib penciling
    - kyphosis;
    - vertebral body scalloping
    - dural ectasia
    - soft tissue masses;

- Idiopathic Tipe M 11YRS

Rizzoli Ortopedic Institute

Bologna

Vertebral Surgery Division



- Idiopathic Tipe M 11YRS

-soft tissue masses

M 12YRS

Rizzoli Ortopedic Institute

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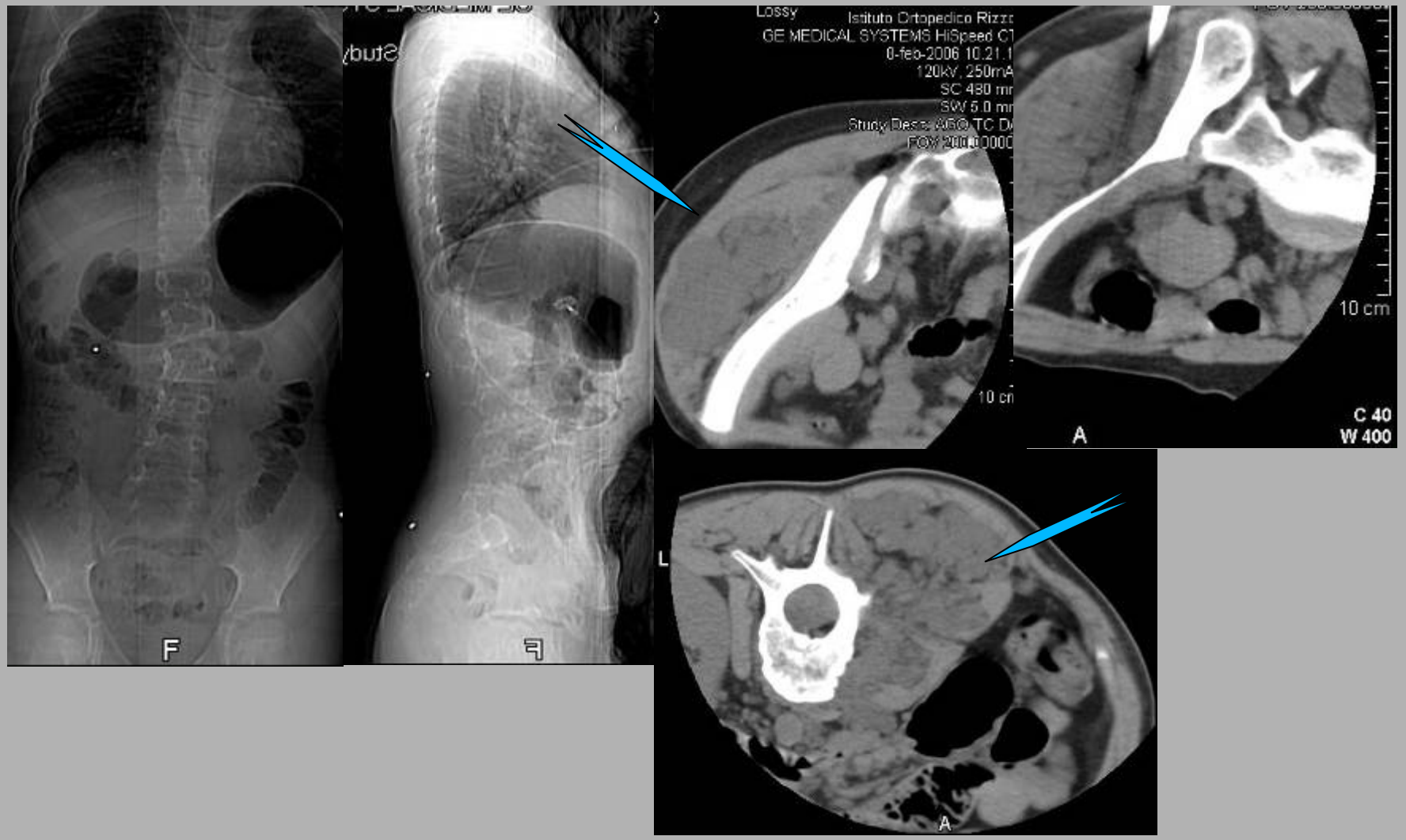
Vertebral Surgery Division



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Pathological dislocation of the hip in neurofibromatosis: a case report.

Lampasi M, Greggi T, Sudanese A.
Chir Organi Mov. 2008 Apr;91(3):163-6.
Epub 2008 May 21.

Surgical Treatment of Scoliosis in Neurofibromatosis

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Vertebral Surgery Division

Spinal Deformity

Nondystrophic type:

Treat like idiopathic scoliosis.
Observe curves less than 20 degrees
Brace from 20 to 35 degrees.
Consider posterior spinal fusion with instrumentation for curves 35 to 60 degrees
Consider posterior spinal fusion and ant-posterior with instrumentation for curves > 60 degrees

Surgical Treatment of Scoliosis in Neurofibromatosis

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Vertebral Surgery Division

Spinal Deformity

Dystrophic type:

Bracing is ineffective.
If kyphosis less than 49 degrees, consider posterior fusion with instrumentation.
For increased kyphosis or scoliosis greater than 40 degrees, consider anterior-posterior fusion with anterior strut grafting with rib, fibula, iliac crest or tibia and posterior instrumentation. Graft concave side.
Curves less than 20 degrees observe every 6 months.
Curves from 20-40 degrees scoliosis consider posterior fusion and instrumentation. Fusion from neutral vertebrae above and below curve.
Post-op immobilization with Risser cast or TLSO recommended up to one year or until fusion complete. Laminectomy alone is not recommended . Does not eliminate neurological symptoms and results in worsening kyphosis.

Surgical Treatment of Scoliosis in Neurofibromatosis

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Vertebral Surgery Division

Neurologic Deficit Secondary to Spinal Cord Compression

Differential Diagnosis

Intraspinal Lesions
-Neurofibroma
-Shwannoma
-Neuroectodermal Tumors
Secondary Compression
- Kyfosis
- Scoliosis -

Surgical Treatment of Scoliosis in Neurofibromatosis

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TC- MRI: evaluates dystrophic forms to characterize soft tissue masses;

Treatment: of Dysplastic Scoliosis;

- one can expect relentless progression that is refractory to bracing;
- surgical fusion is therefore required;

- scoliosis alone: posterior arthrodesis (w/ internal fixation);

- associatted kyphosis

there is increases risk of paraplegia & pseudo-arthrosis following surgery;
anterior fusion combined w/ posteiror instrumentation & fusion is indicated
when kyphosis > 50 deg or scoliosis is > 80 deg;

Surgical Treatment of Scoliosis in Neurofibromatosis

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-Winter, R.B., Moe, J.H., Bradford, D.S., Lonstein, J.E., Pedras, C.V., Weber, A.H. Spine Deformity in Neurofibromatosis. JBJS 1979;61-A: 677-694

-BA Akbarnia, KR Gabriel, E Beckman, D Chalk. Prevalence of scoliosisi in neurofibromatosis Spine, 1992 -

- vertebral body scalloping
- dural ectasia

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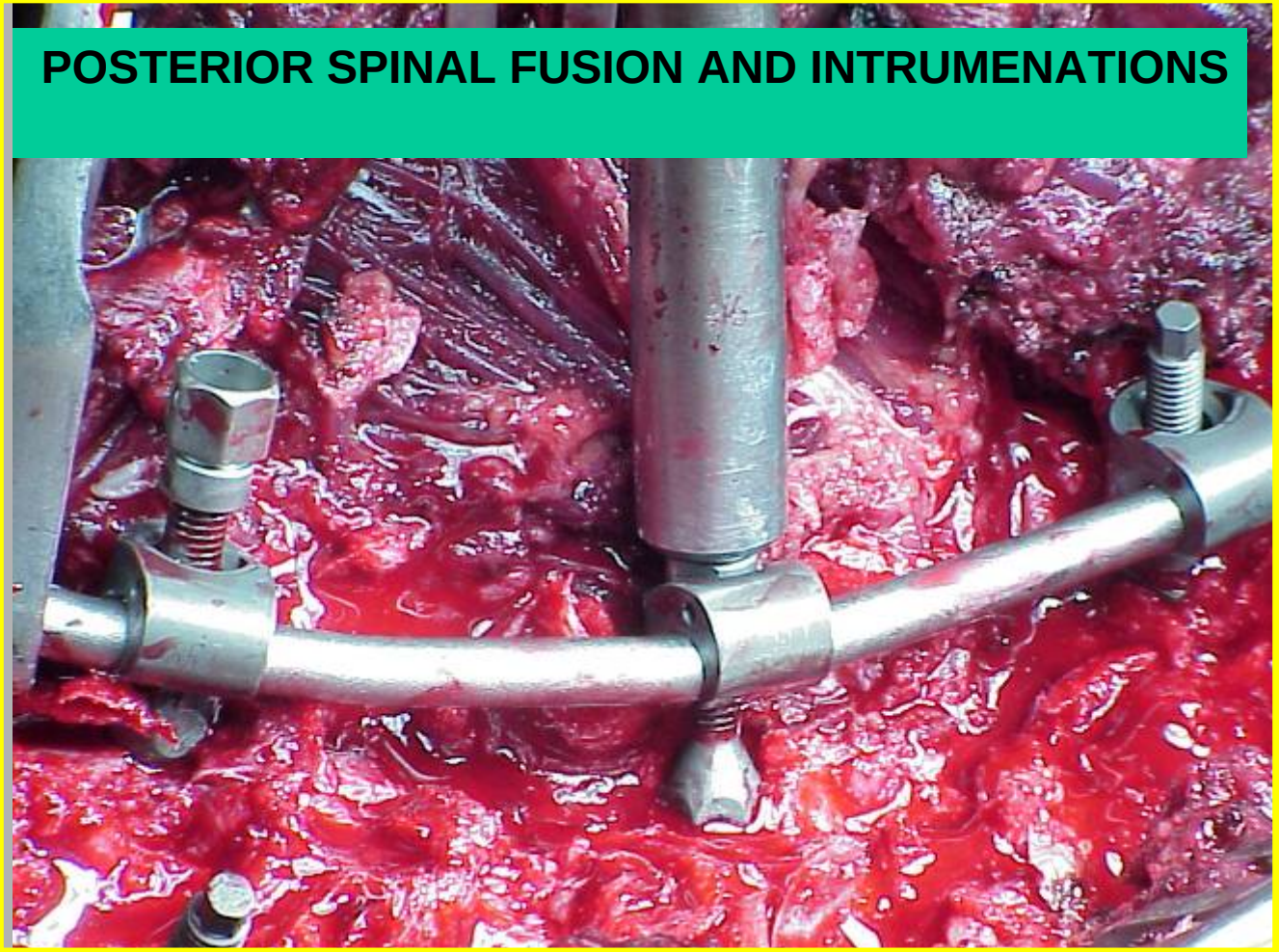
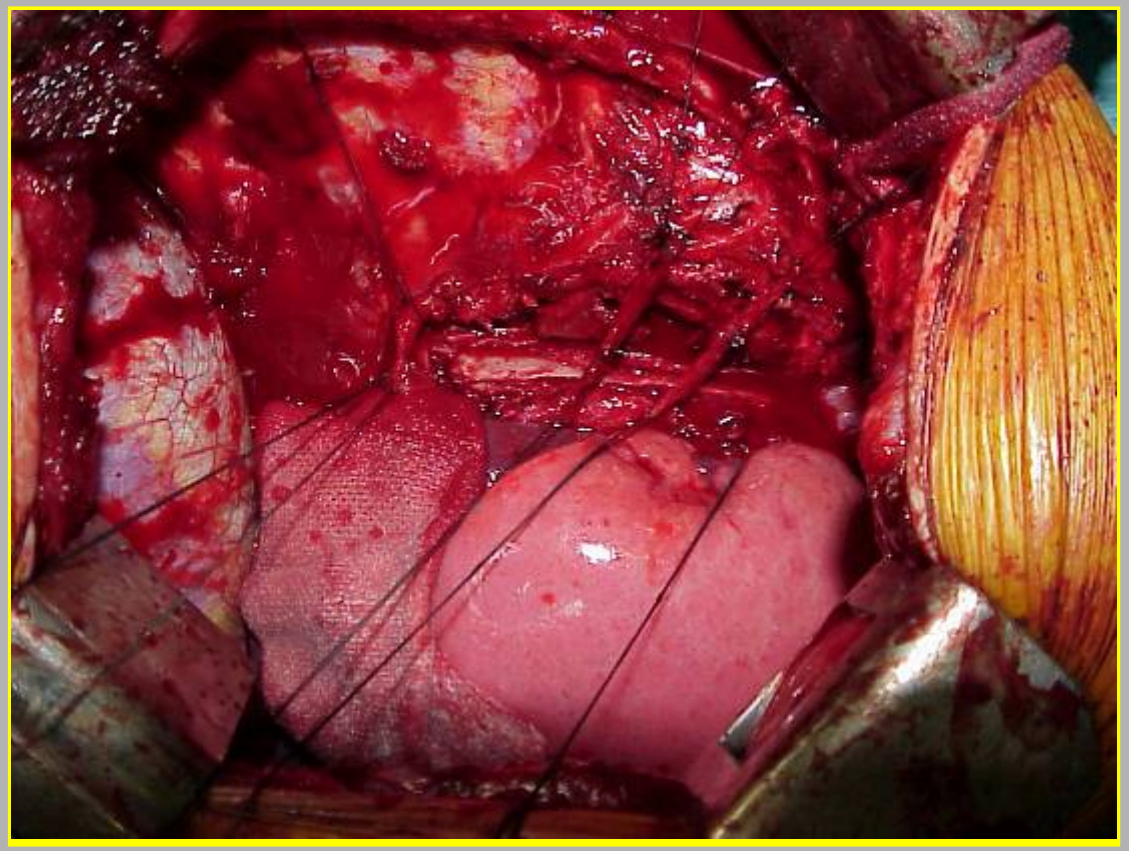
Vertebral Surgery Division

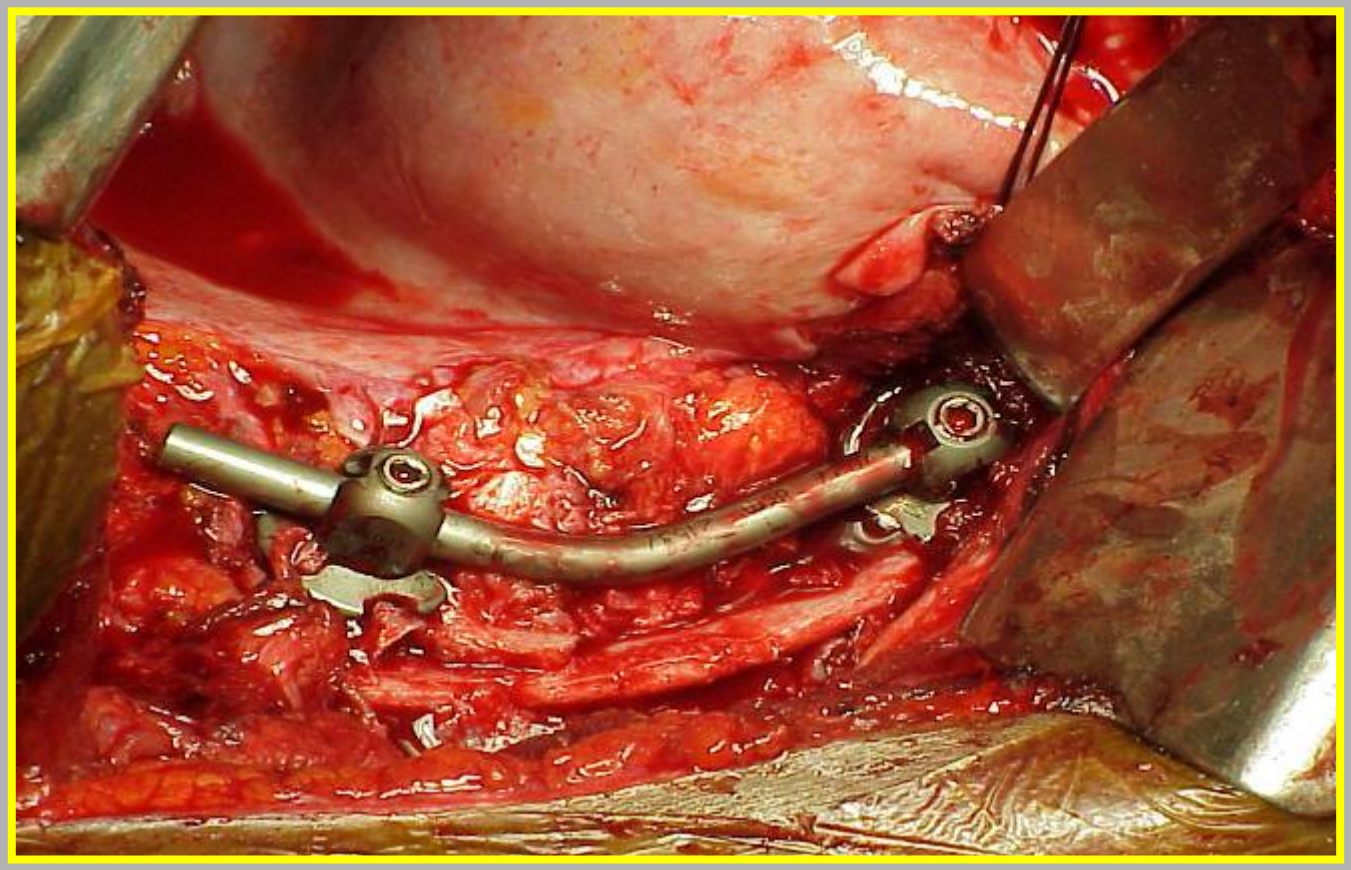
POSTERIOR SPINAL FUSION AND INTRUMENATIONS



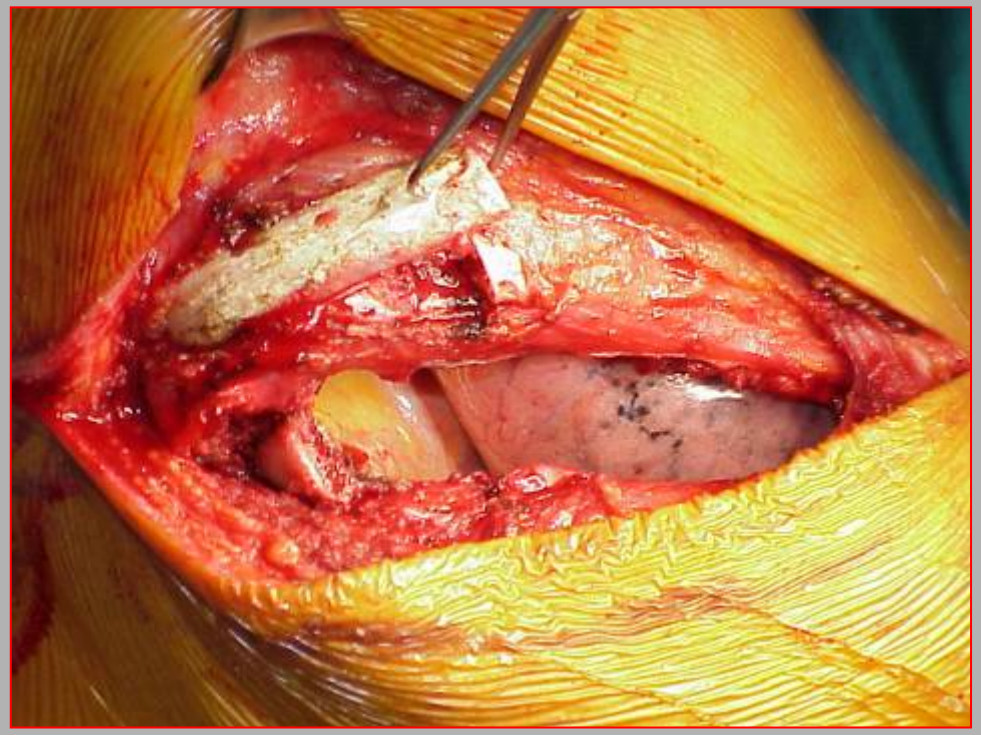
Posterior instrumentations

ANTERIOR ARTRODESIS TORACOTOMIA

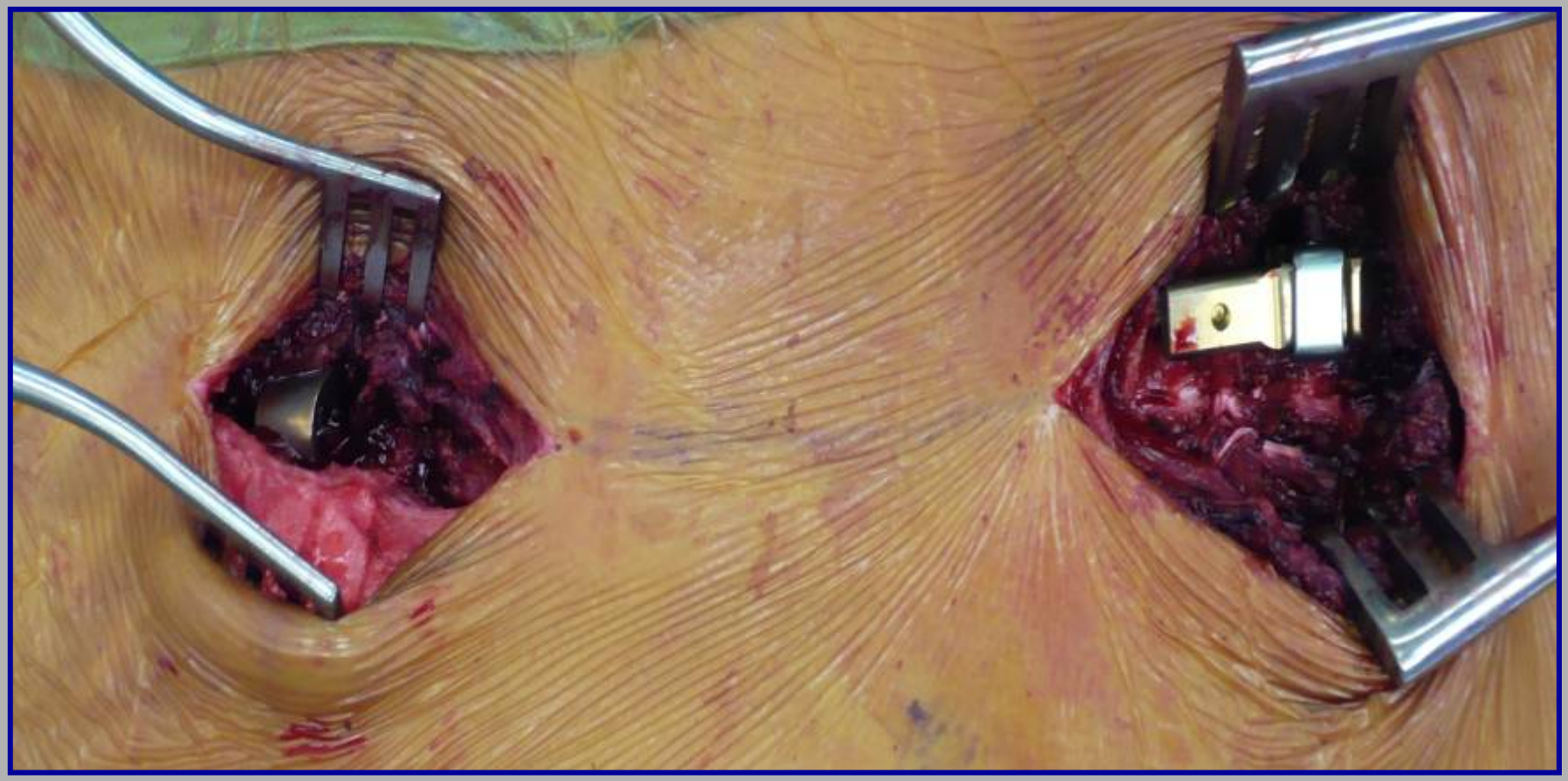




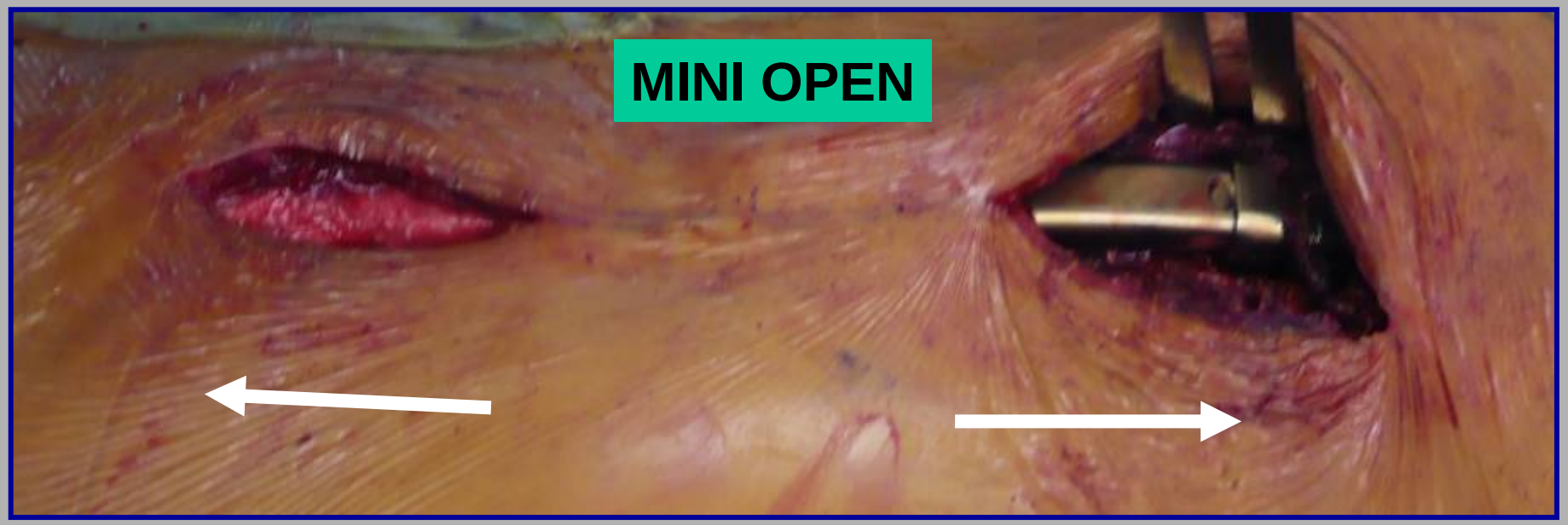
ANTERIOR INSTRUMENTATION

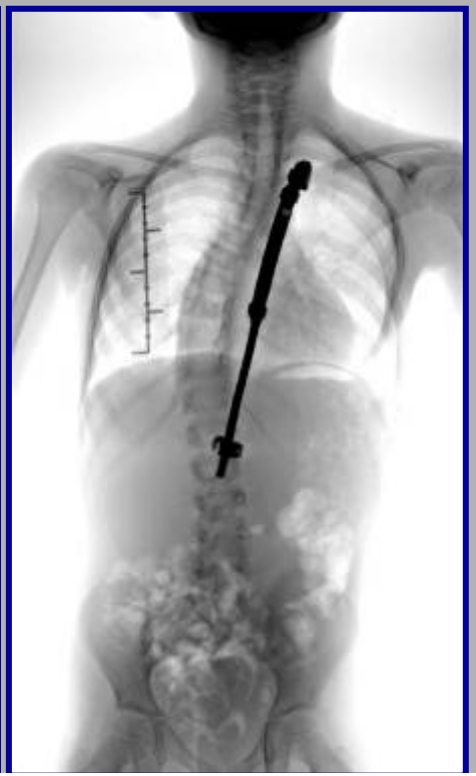


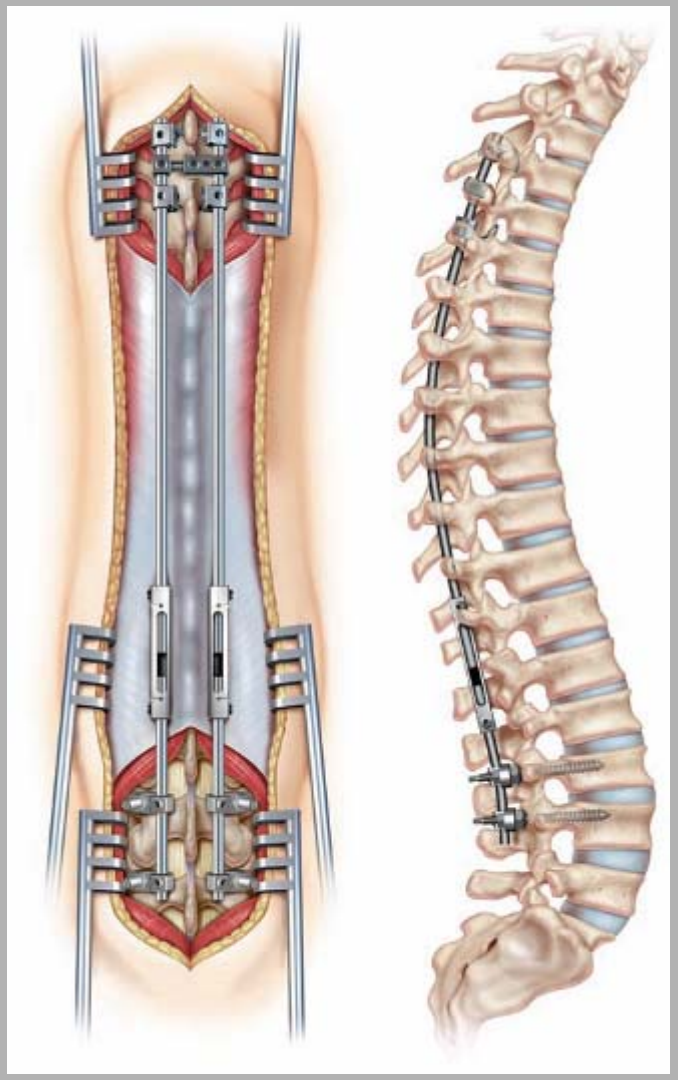
“HALO TRACTION”



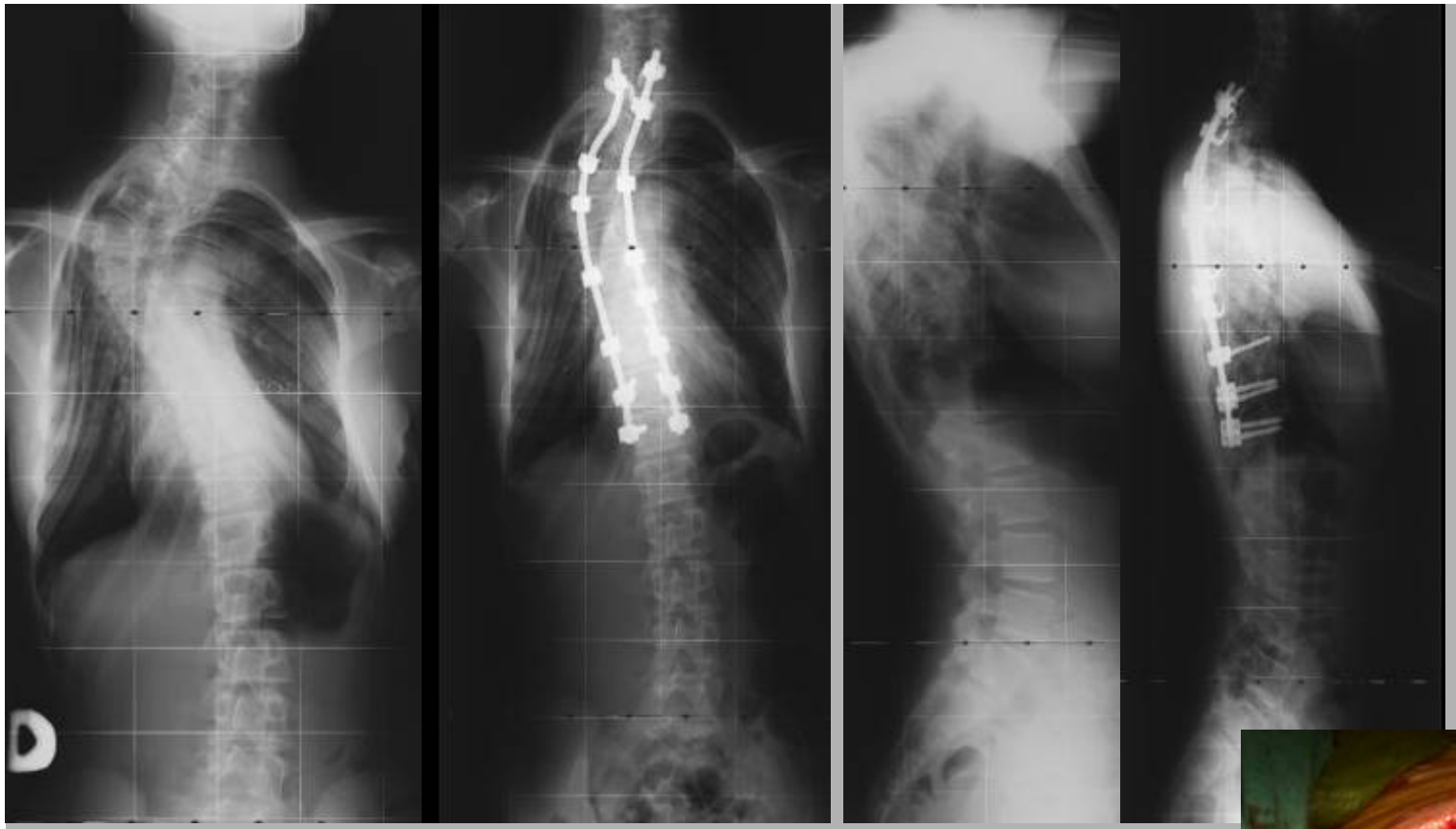
GROWING-SPINE


MINI OPEN

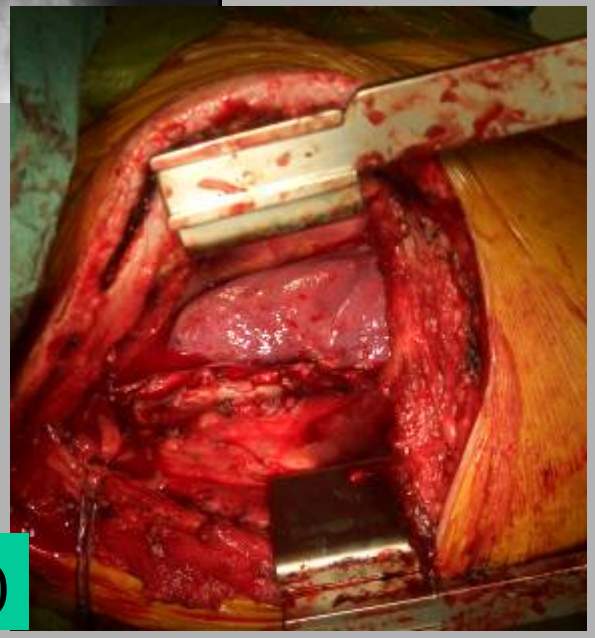




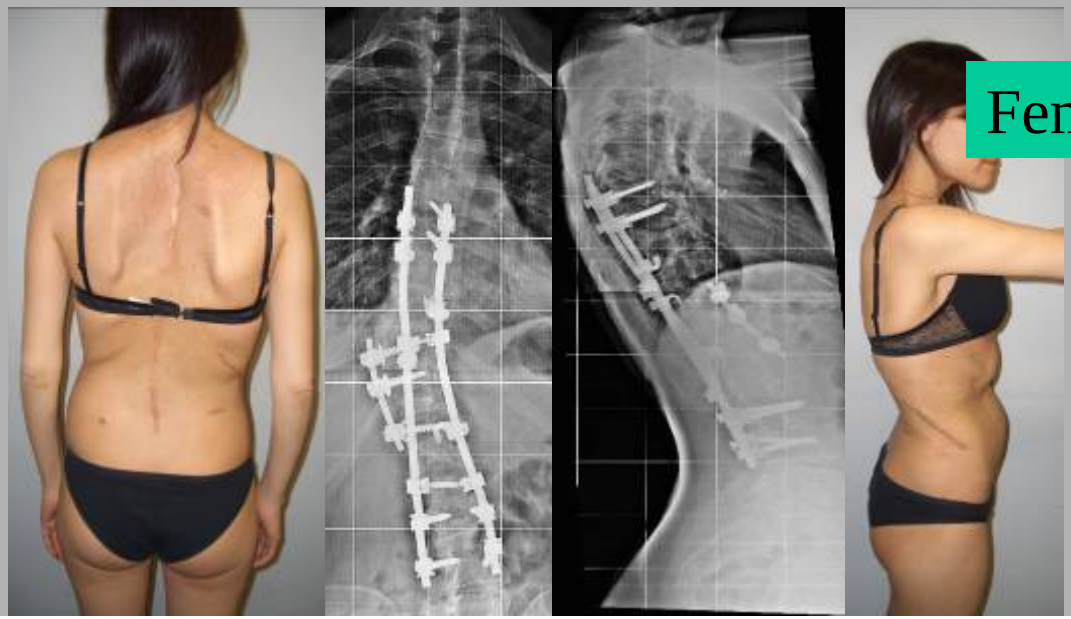
Dual rod



Female 13 years-old (NF-1)

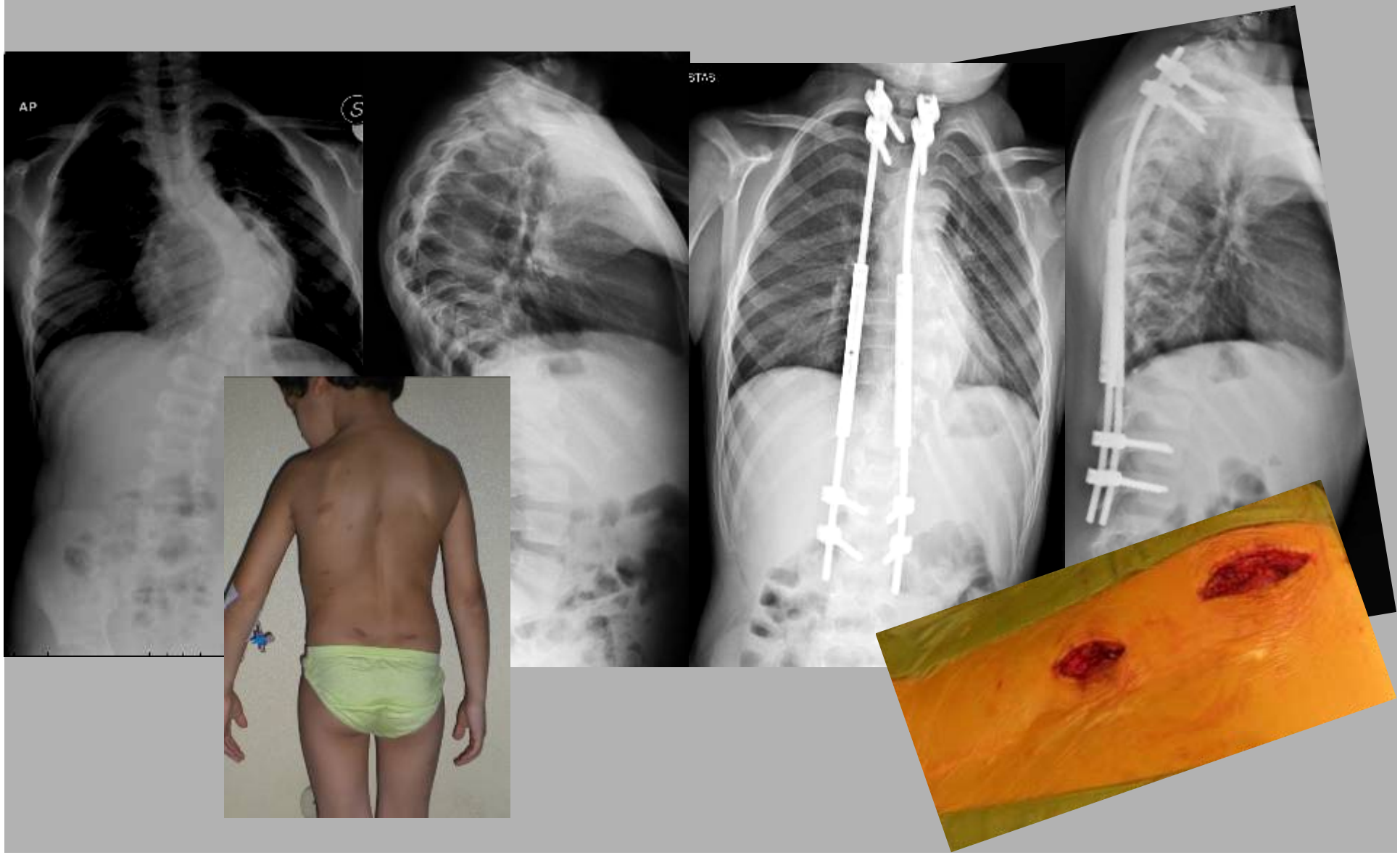


Polson’s Thoracotomy)

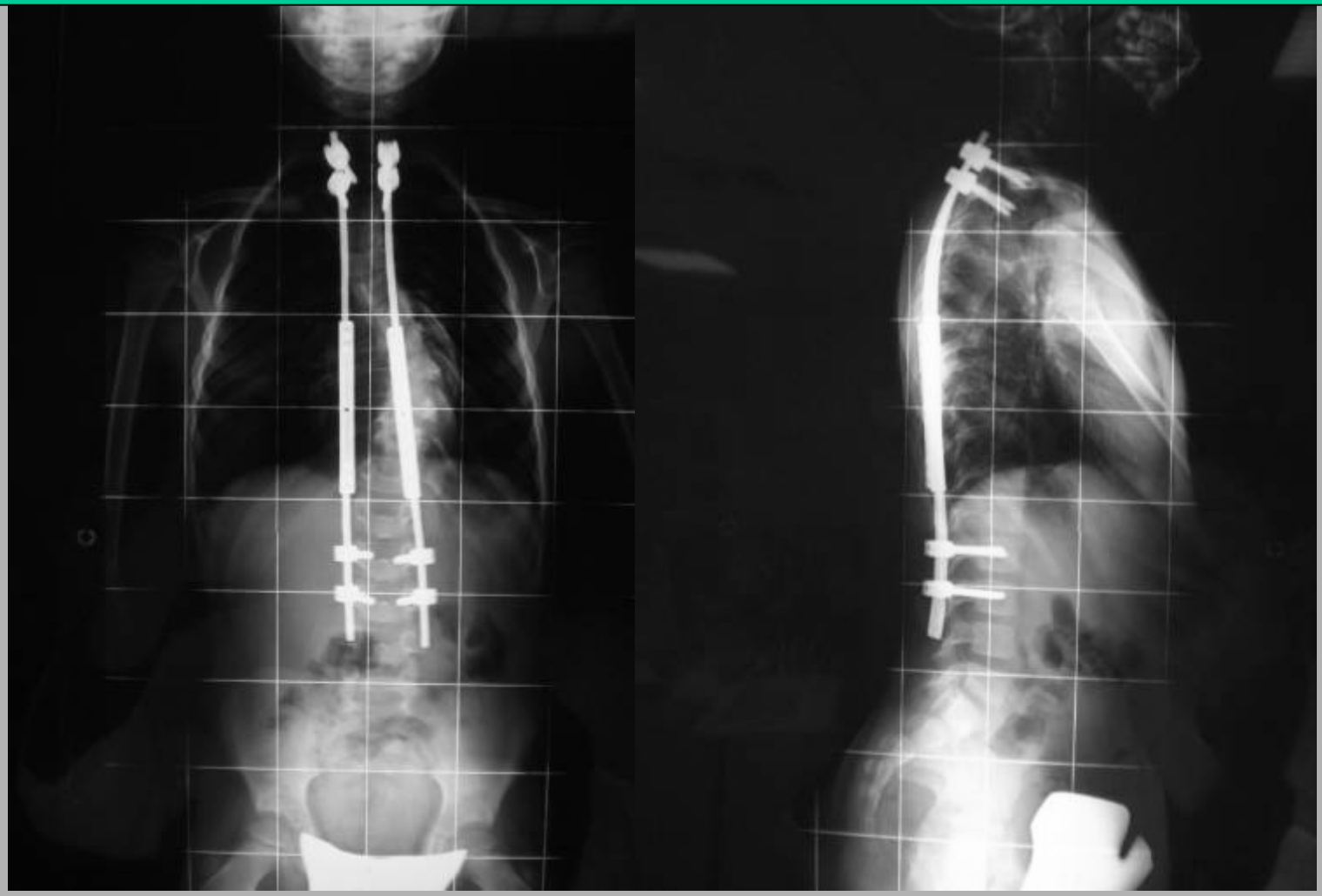


Female 28 years-old (NF-1)

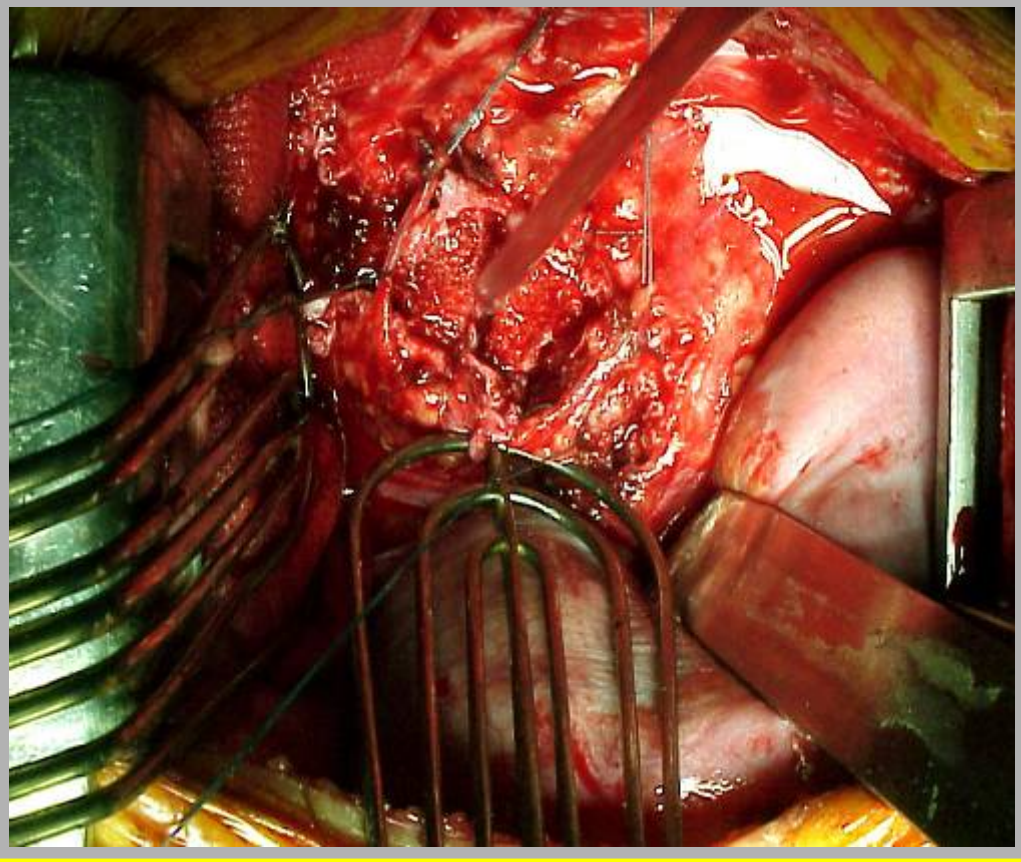
Male 7 years-old (NF-1)



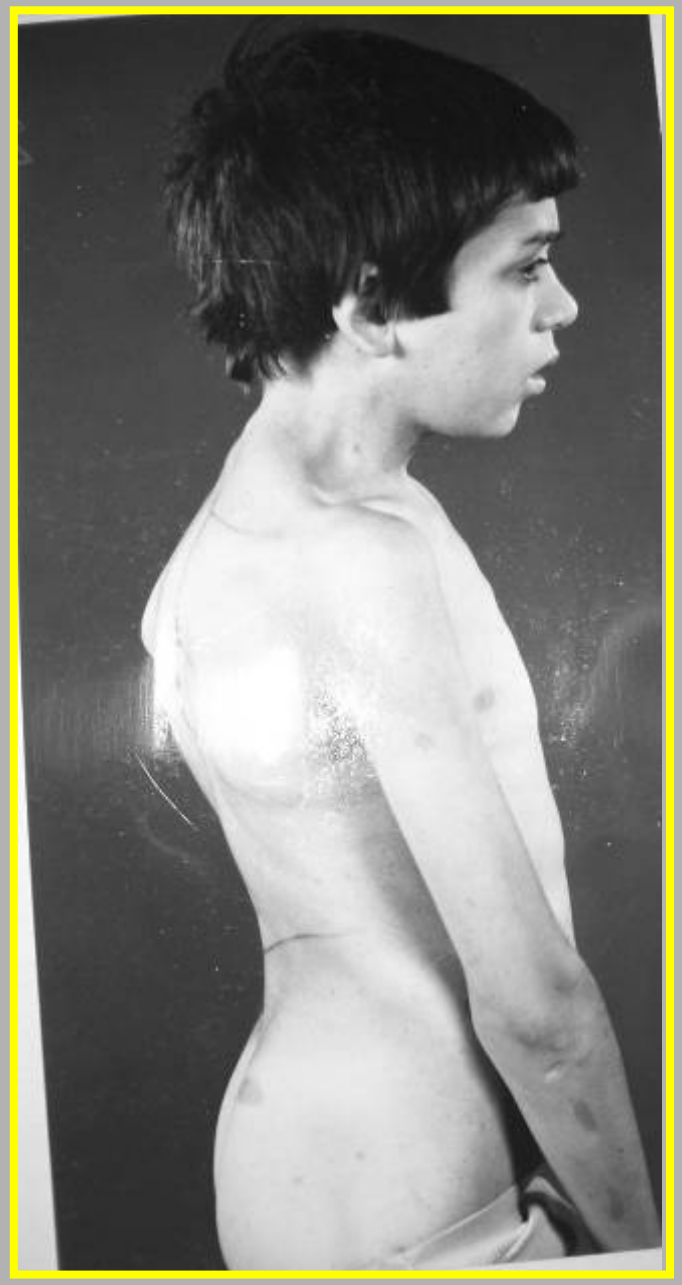
Male 7 years-old (NF-1)

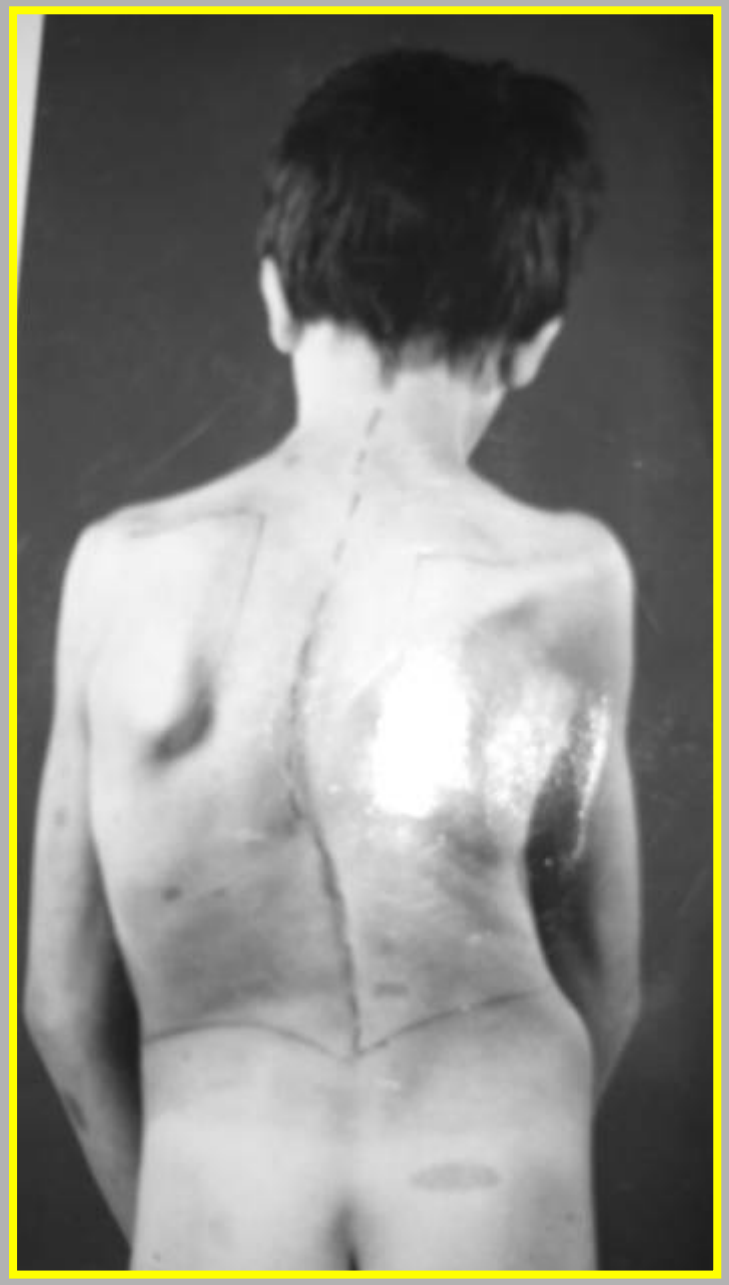


ARTHRODESIS BY DOUBLE ACCESS



SPINE fusion
BY ANTERIOR AND POSTERIOR TIME



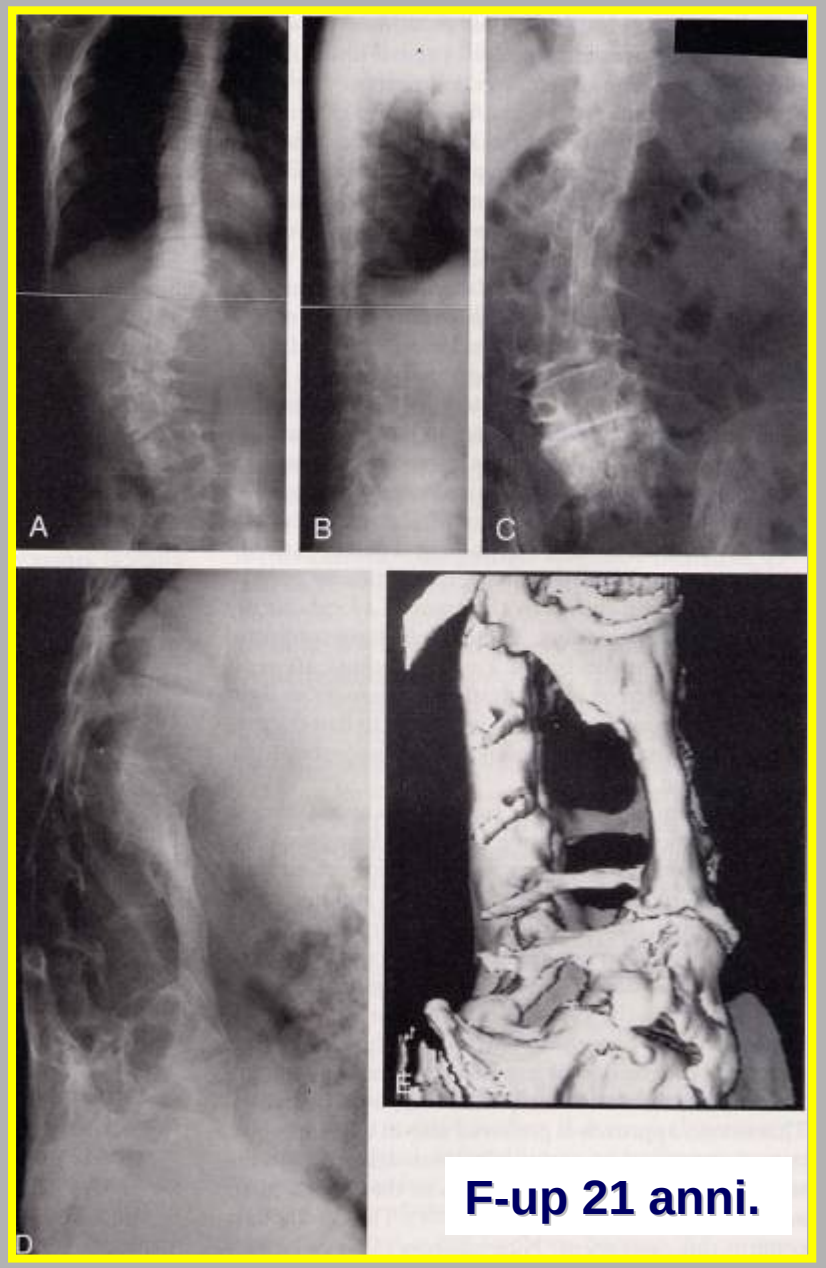


M. 12 yrs DYSTRPHIC SCOLIOSIS in NEUROFIBROMATOSIS

M. 42 yrs

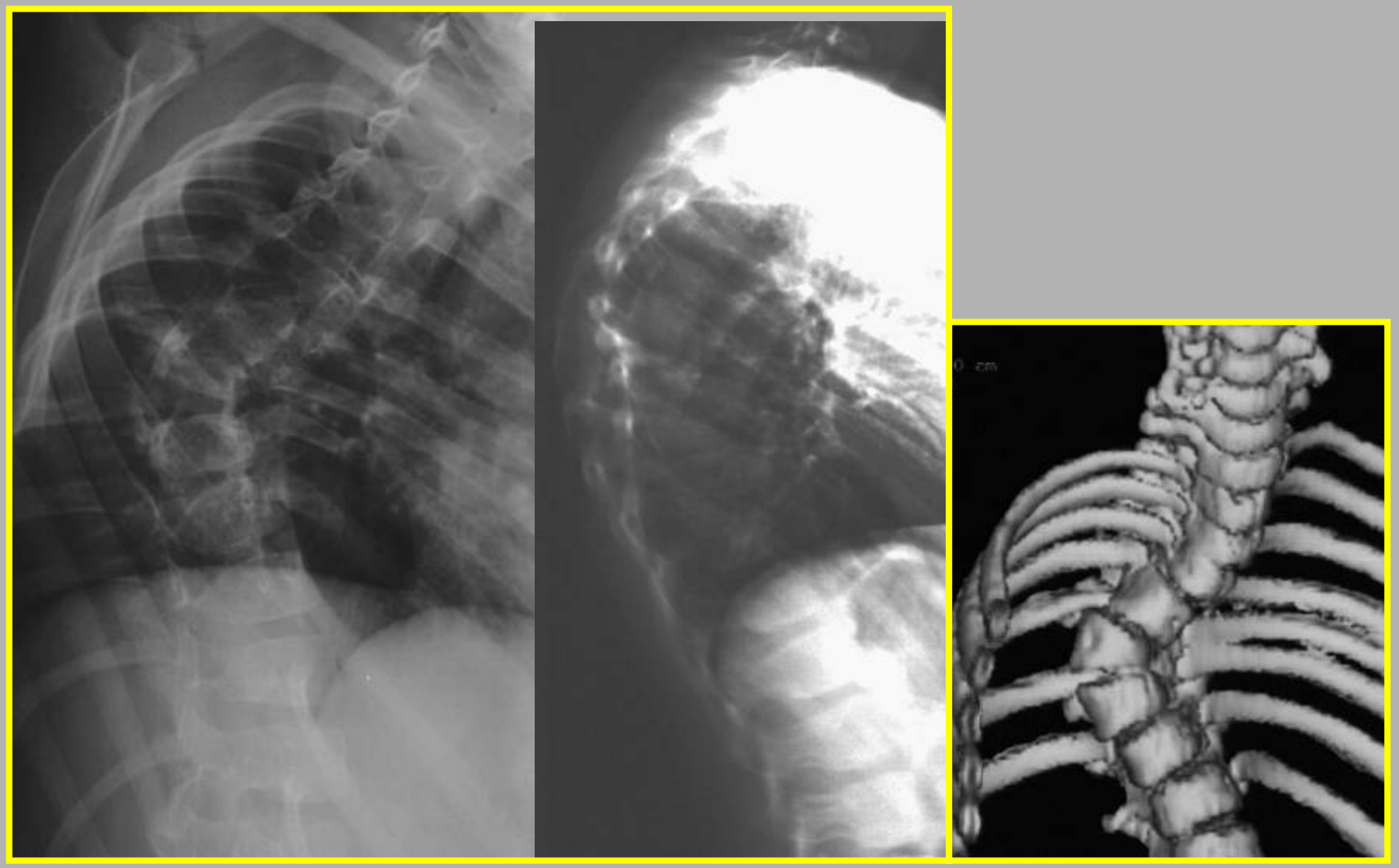
2007

30yrs F-up

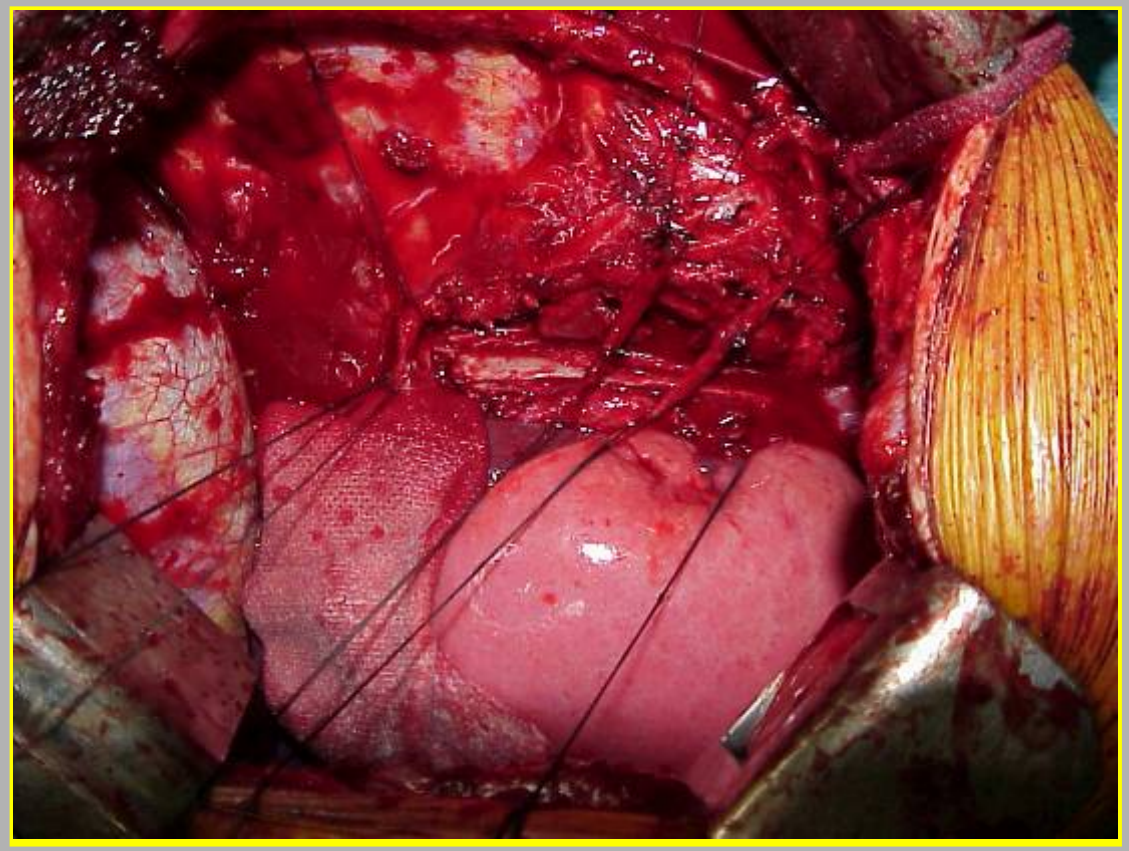


M. 13 SCOLIOSIS ORIGINATED BY NEUROFIBROMATOSIS

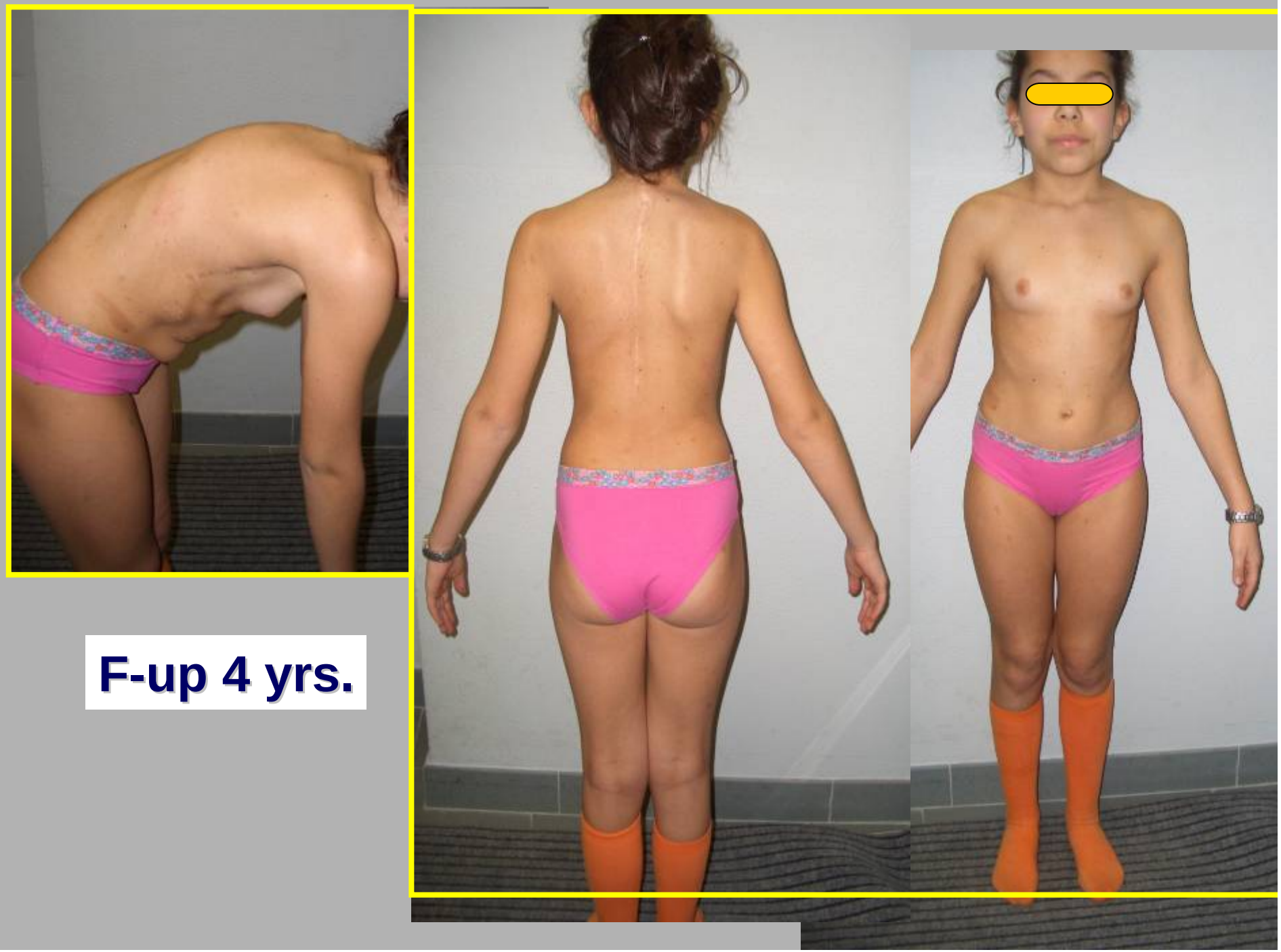
F.,5 CIFOSCOLIOSIS NF-1 dystrophic deformity



Anterior spine arthrodesis : using toracotomy

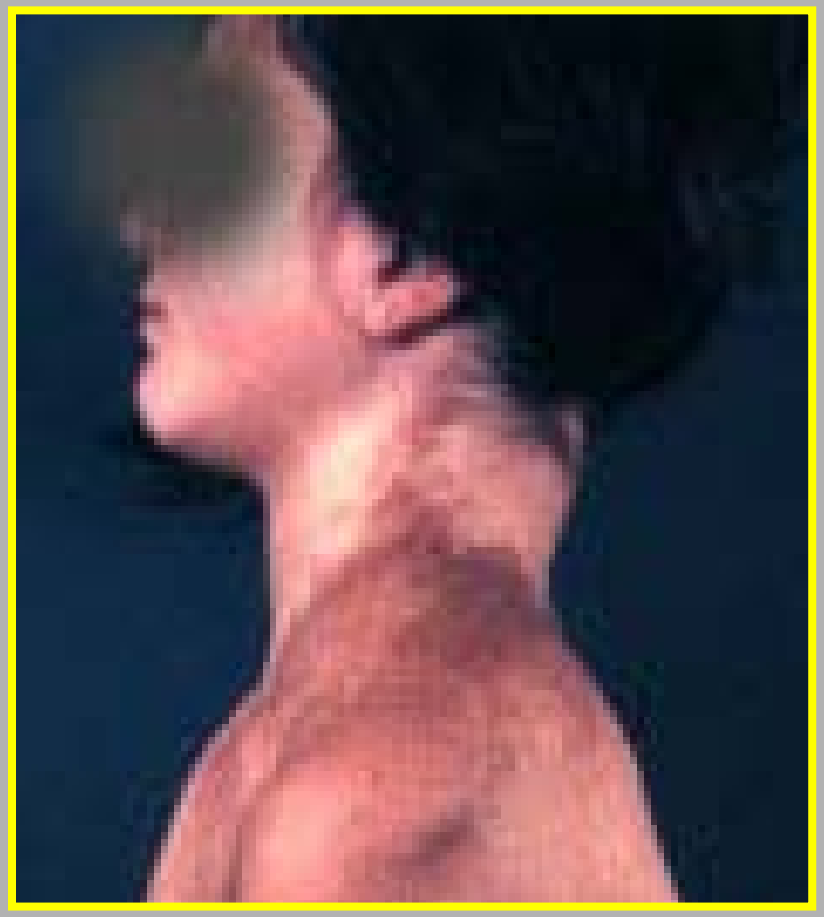


F-UP 2 yrs.



F-up 4 yrs.

F 8 yrs.
CERVICAL KHYPHOSIS
originated by
NEUROFIBROMATOSIS



F-UP Rx 10 yrs.

Complex cases

Diagnostic difficulty

Careful study and Clinical observation

Families' collaboration and involvement

Importance of precocious diagnosis:

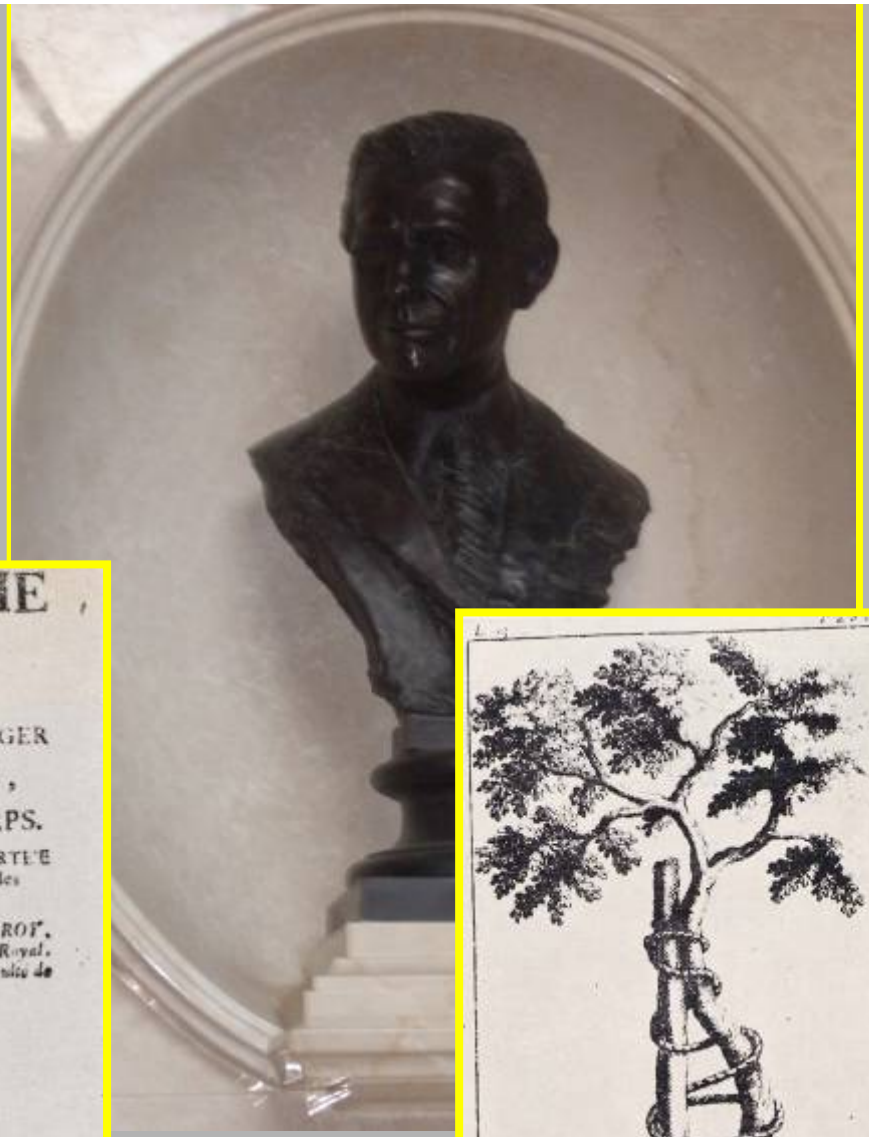
the role of Genetic

Availability of multidisciplinary expertise

Better use of technical and available organizational resources

Guarantee the assistance of the best quality

Continuous updating by a National and International Research Network



L'ORTHOPEDIE,

OU

L'ART

DE PREVENIR ET DE CORRIGER

DANS LES ENFANS,

LES DIFFORMITES DU CORPS.

LE TOUT PAR DES MOYENS A LA PORTE'E des Peres & des Meres, & de toutes les Personnes qui ont des Enfans à élever.

PAR M. ANDRY, CONSEILLER DU ROY, Lecteur & Professeur en Medecine au College Royal, Docteur-Regent, & ancien Doyen de la Faculté de Medecine de Paris, &c.

Avec Figures.

TOME PREMIER.

A PARIS, RUE SAINT JACQUES.

Chez { La Veuve ALIX, au-dessus de la rue des Noyers, au Griffon. / LAMBERT & DURAND, à la Sagesse, & à Saint Landry.

M. DCC. XLI.

AVEC APPROBATIONS ET PRIVILEGE DU ROY.