Congenital disorders

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Congenital abnormalities

The child is born with them

1. Typical - genetic, chromosomes
   and genes abnormality,
   familiar occurence

2. Atypical – not genetic
   embryonal damage
   during pregnancy

Gene mutation

Chromosomal disorders

Autosomal dominant transmission
Autosomal recessive transmission

X-linked disorders

Classification

1. Genetic disorders of cartilage and bone growth
Achondroplasia, epiphyseal dysplasia..)

2. Collagen disorders
Osteogenesis imperfecta, Marfan syndrom
Neurofibromatosis ..

3. Enzyme defects
Mucopolysacharidosis, Gaucher disease..

4. Chromosome disorders
Down syndrom ..

Classification

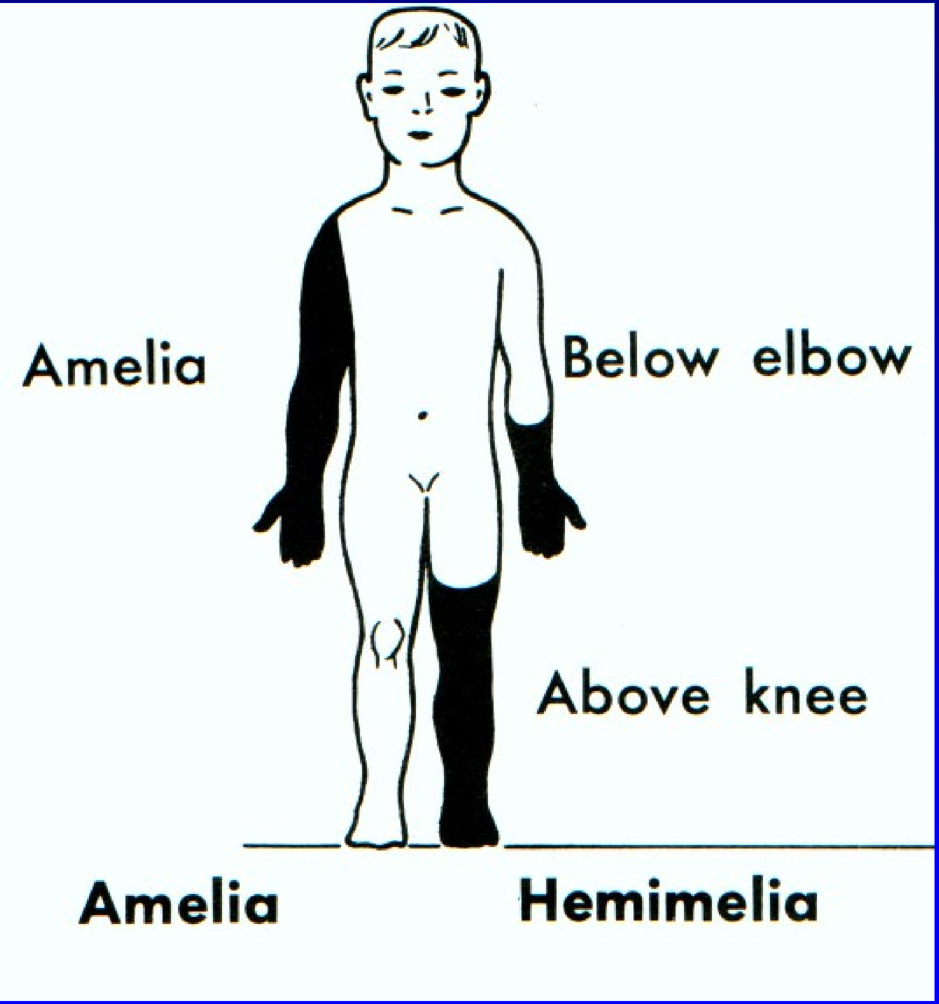
1. Disturbance of a form (shape)
2. Disturbance of separation
3. Duplication
4. Gigantism
5. Hypoplasia
6. Congenital constrictions
7. General deformities

1. Disturbace of a form

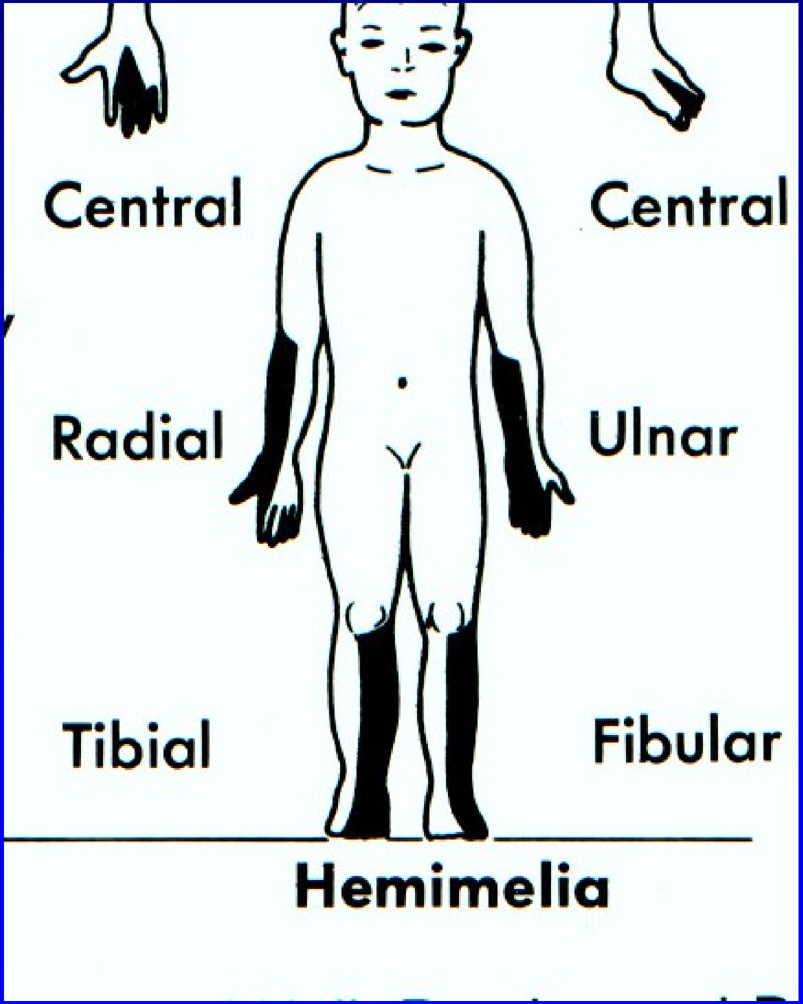
A. Transversal defects
- terminal
- intercalary

B. Longitudinal defects
- terminal
- intercalary

Terminal defects

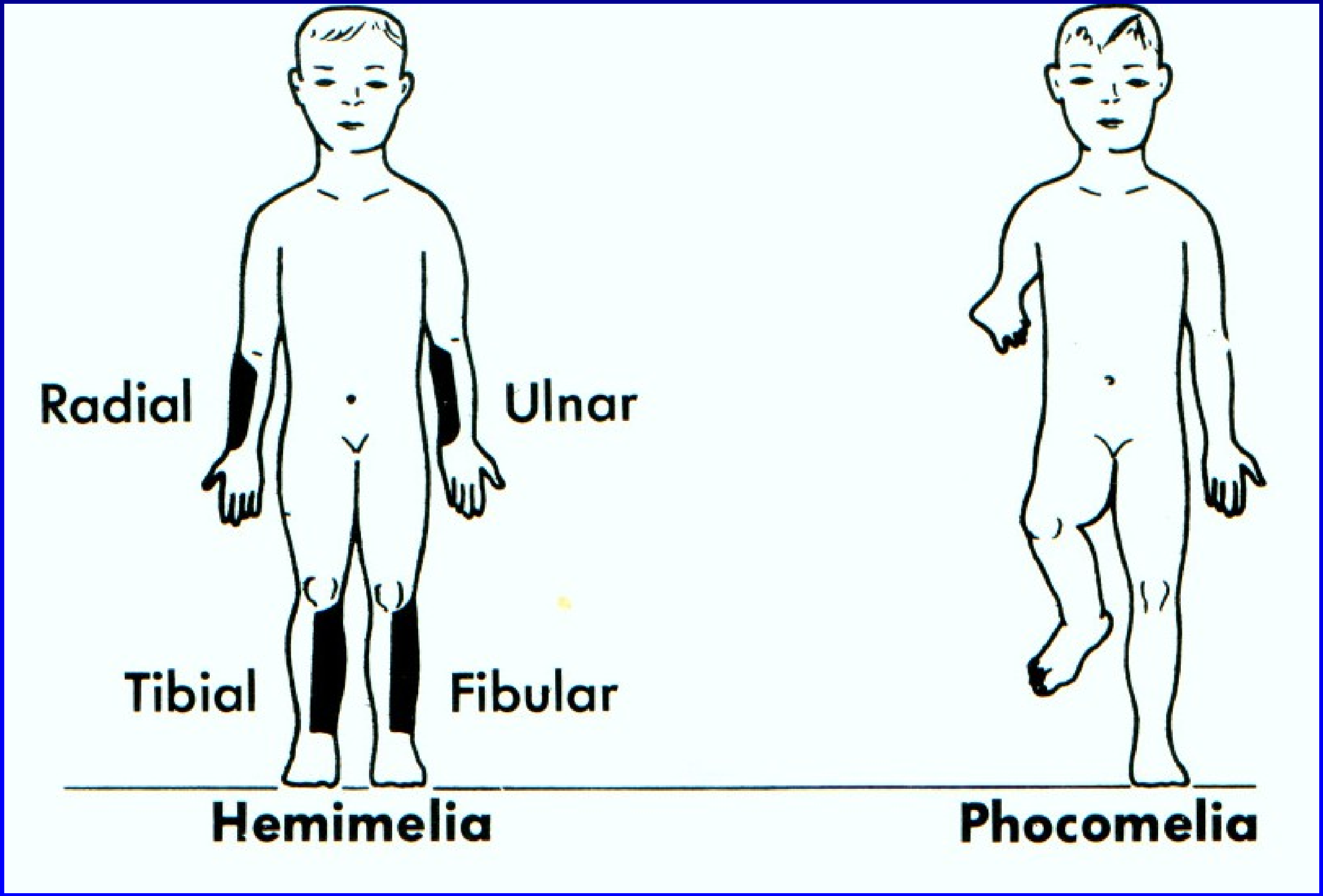


Obr. 1



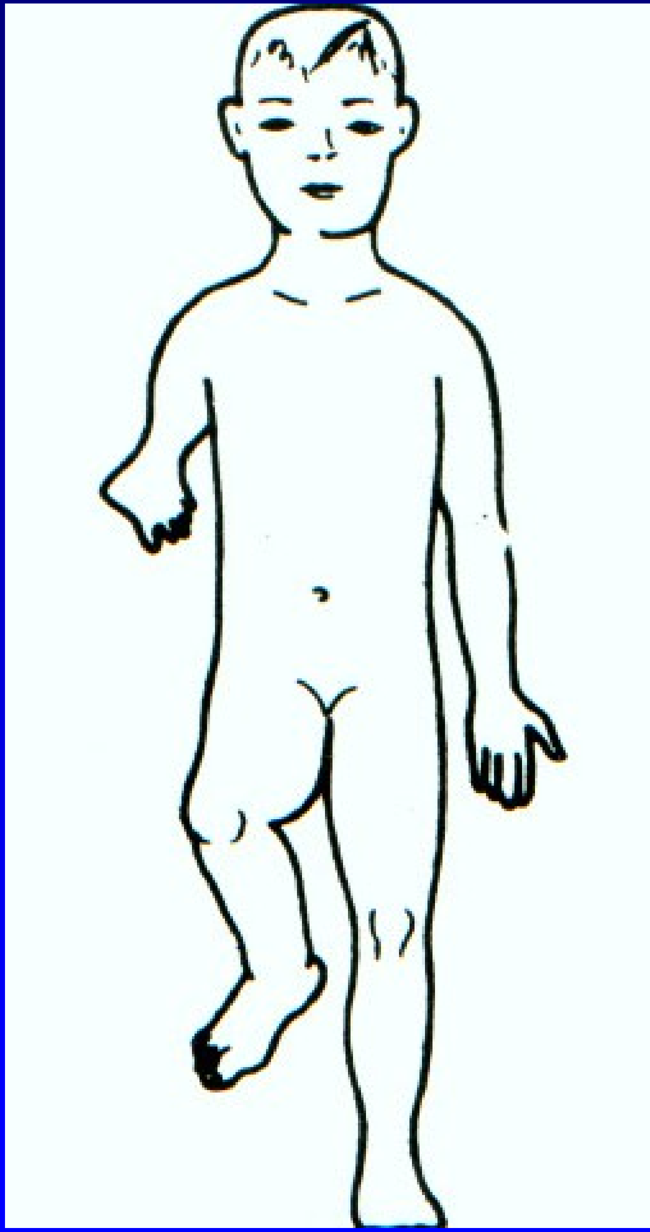
Obr. 2

Intercalary defects



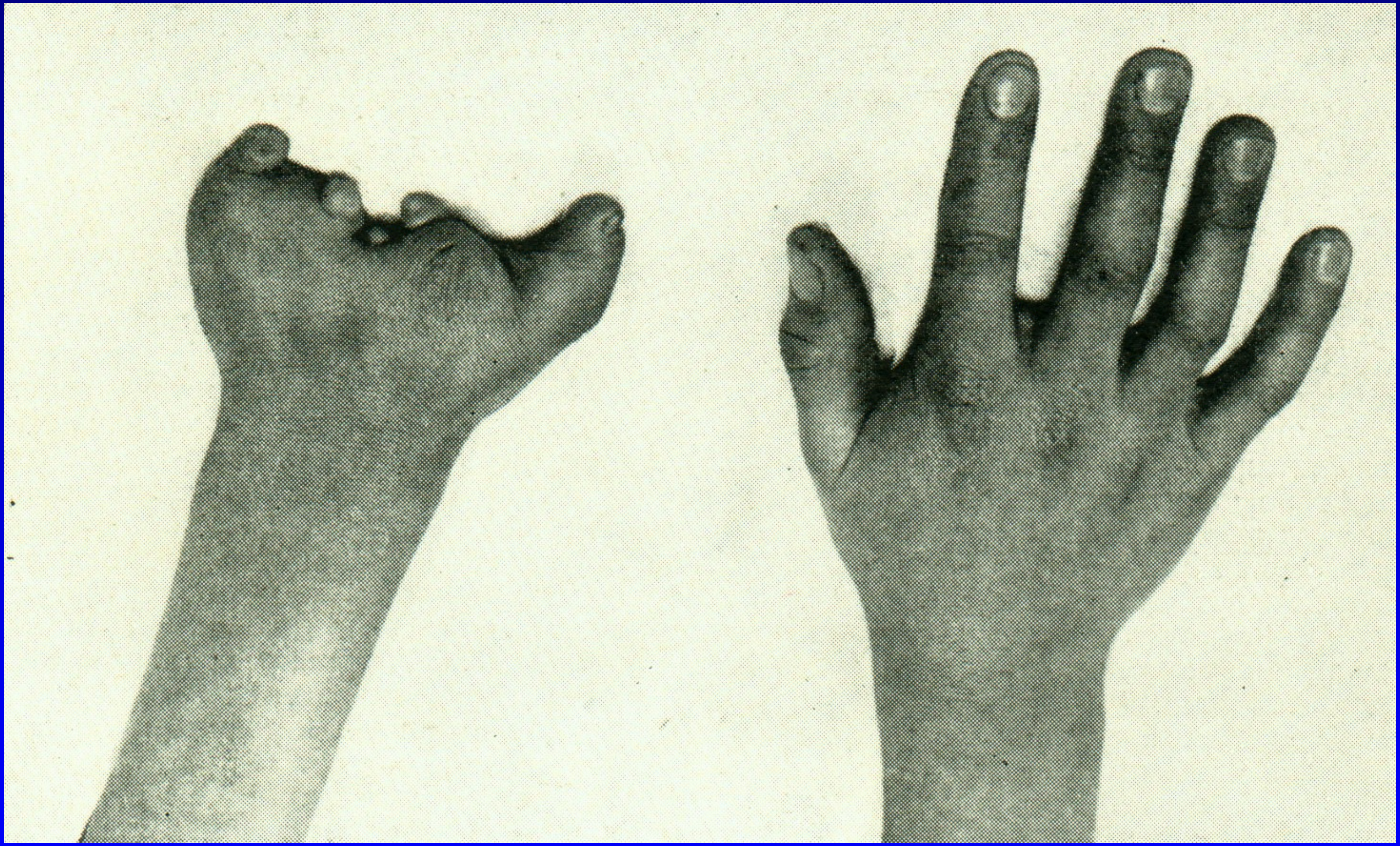
Obr. 3

Focomelia



Obr. 4

Congenital aplasia of fingers



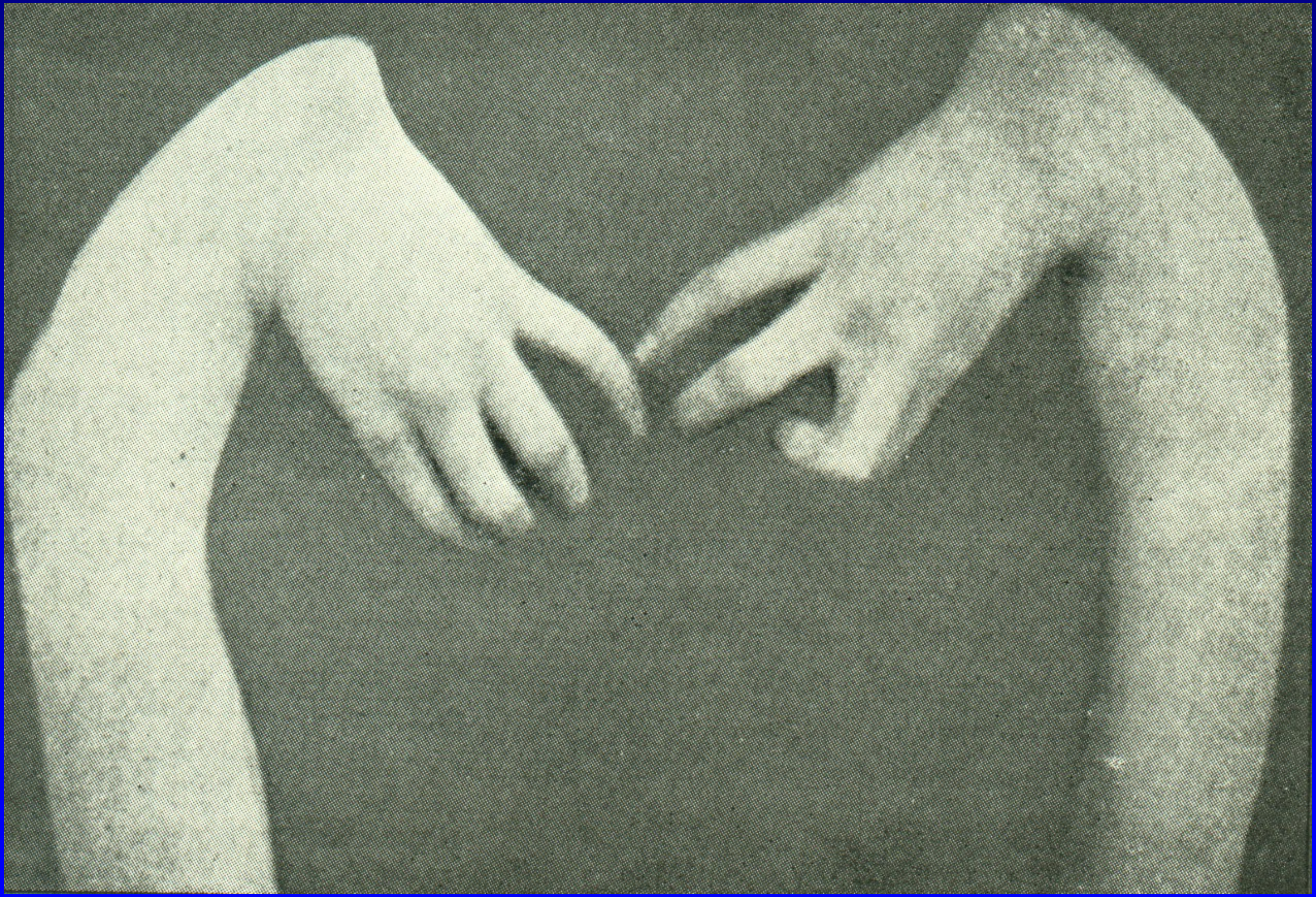
Obr. 5

Dysostosis cleidocranialis

Absence of clavicle

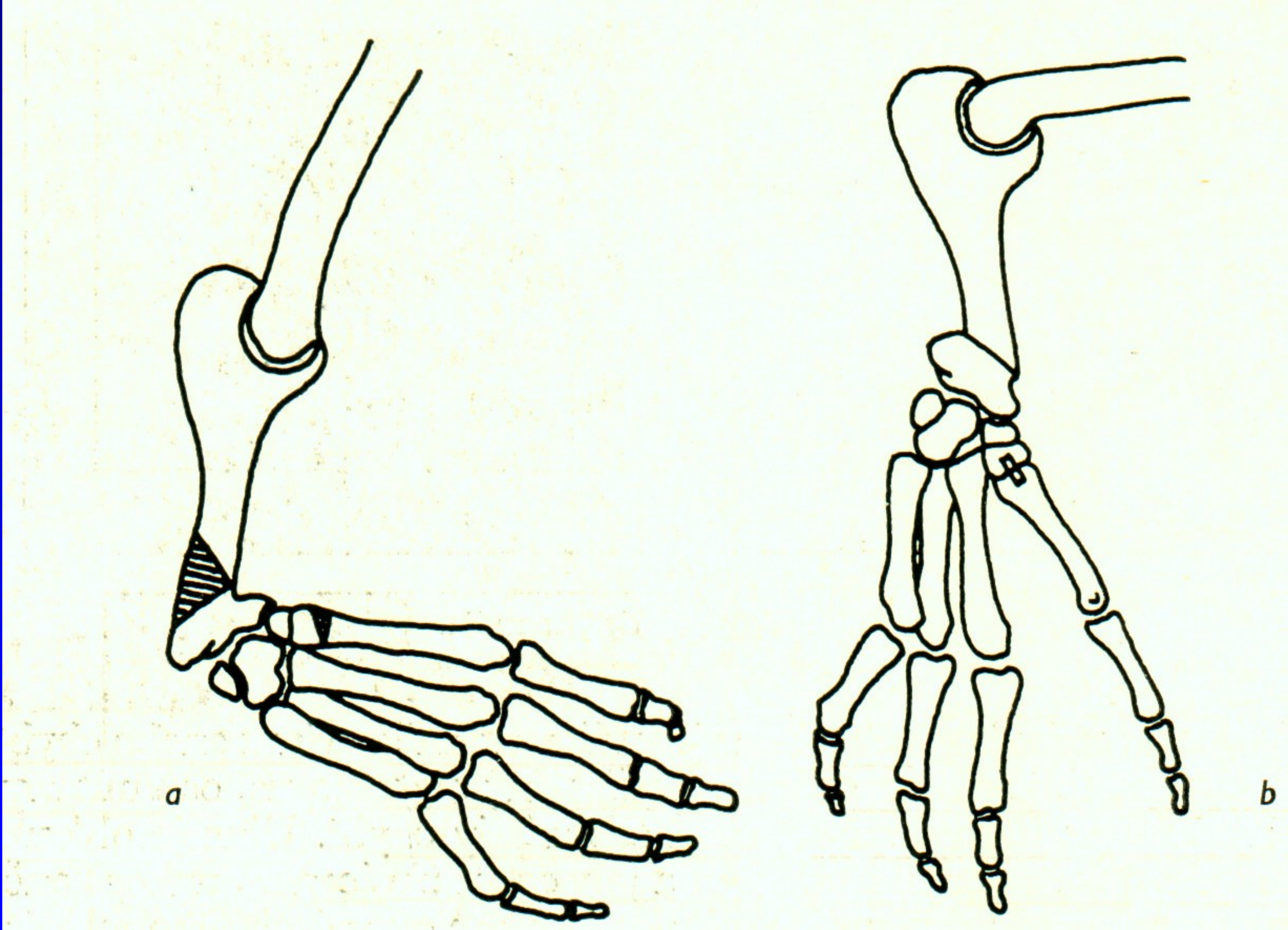
Obr. 6

Congenital aplasia of radius
- manus vara



Obr. 7

Correction surgery for manus vara

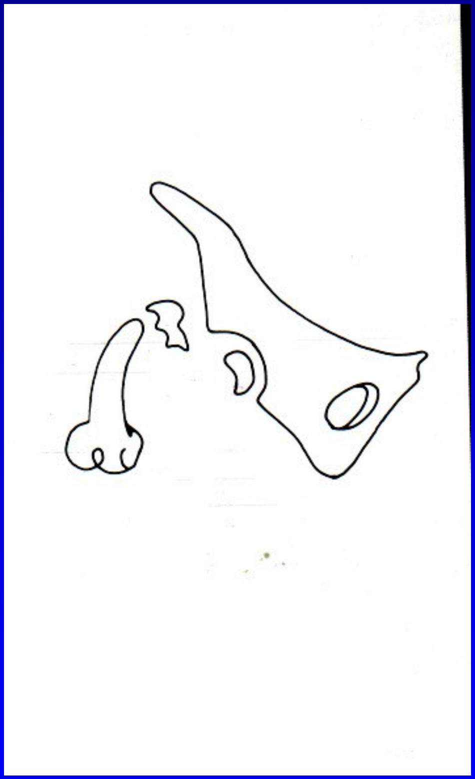


Obr. 8

Cleft hand

Obr. 9

Aplasia of the femur



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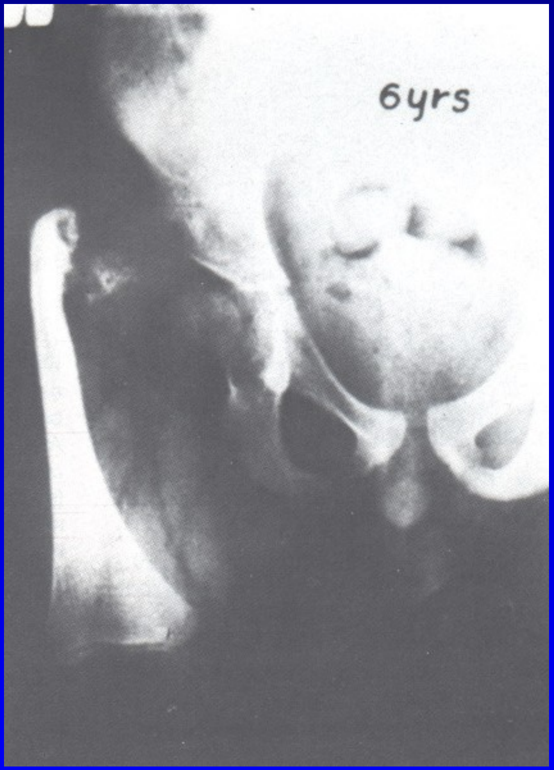
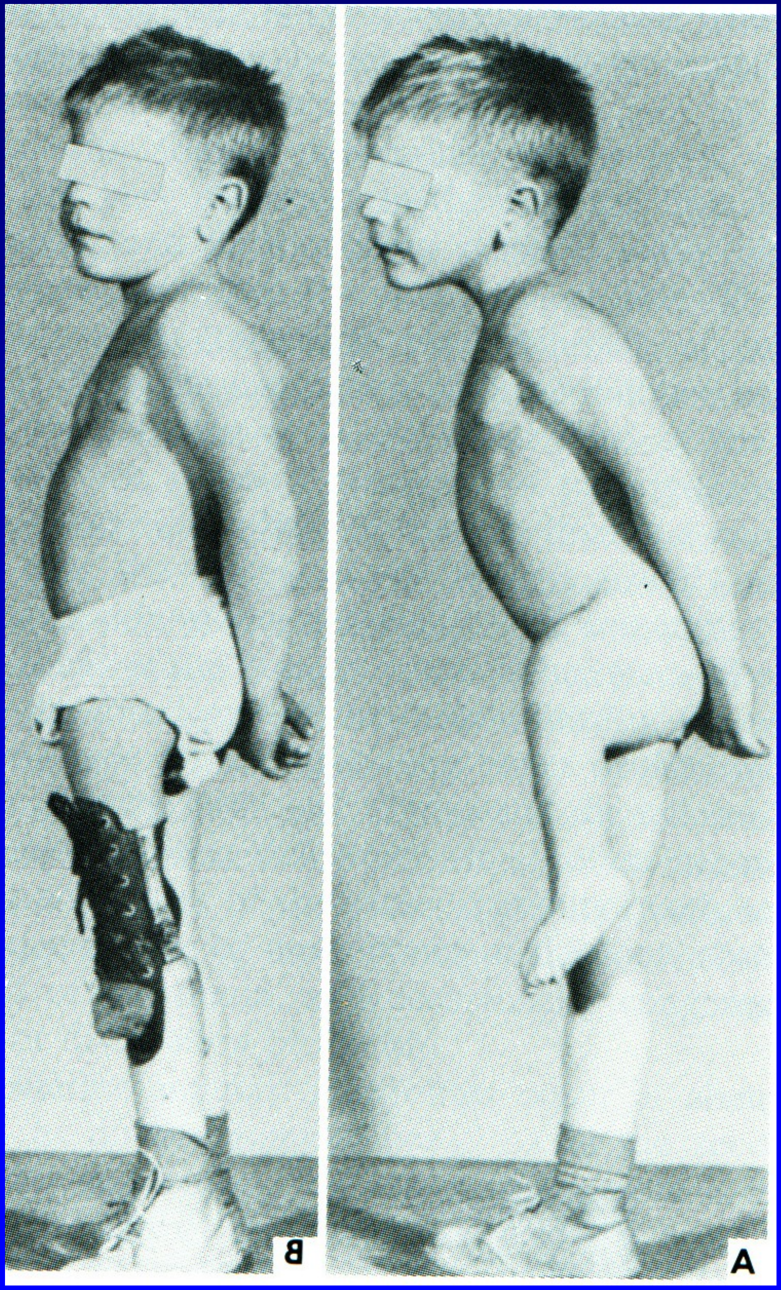


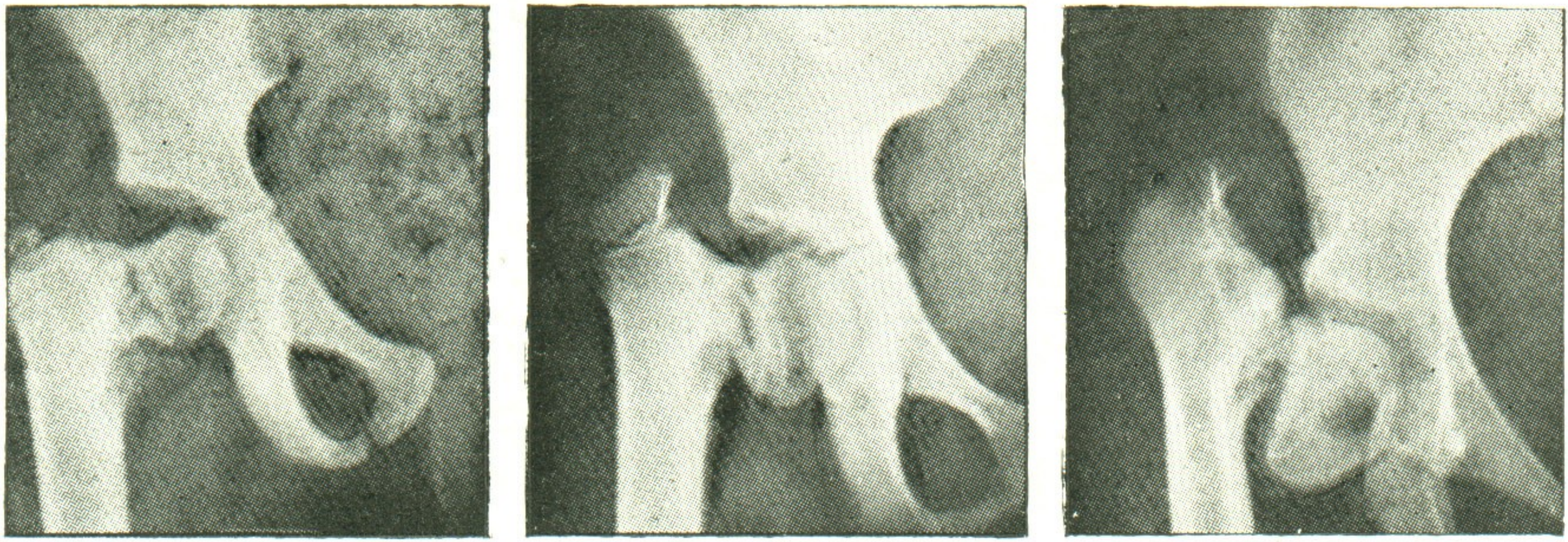
Obr. 11

Congenital aplasia of the femur



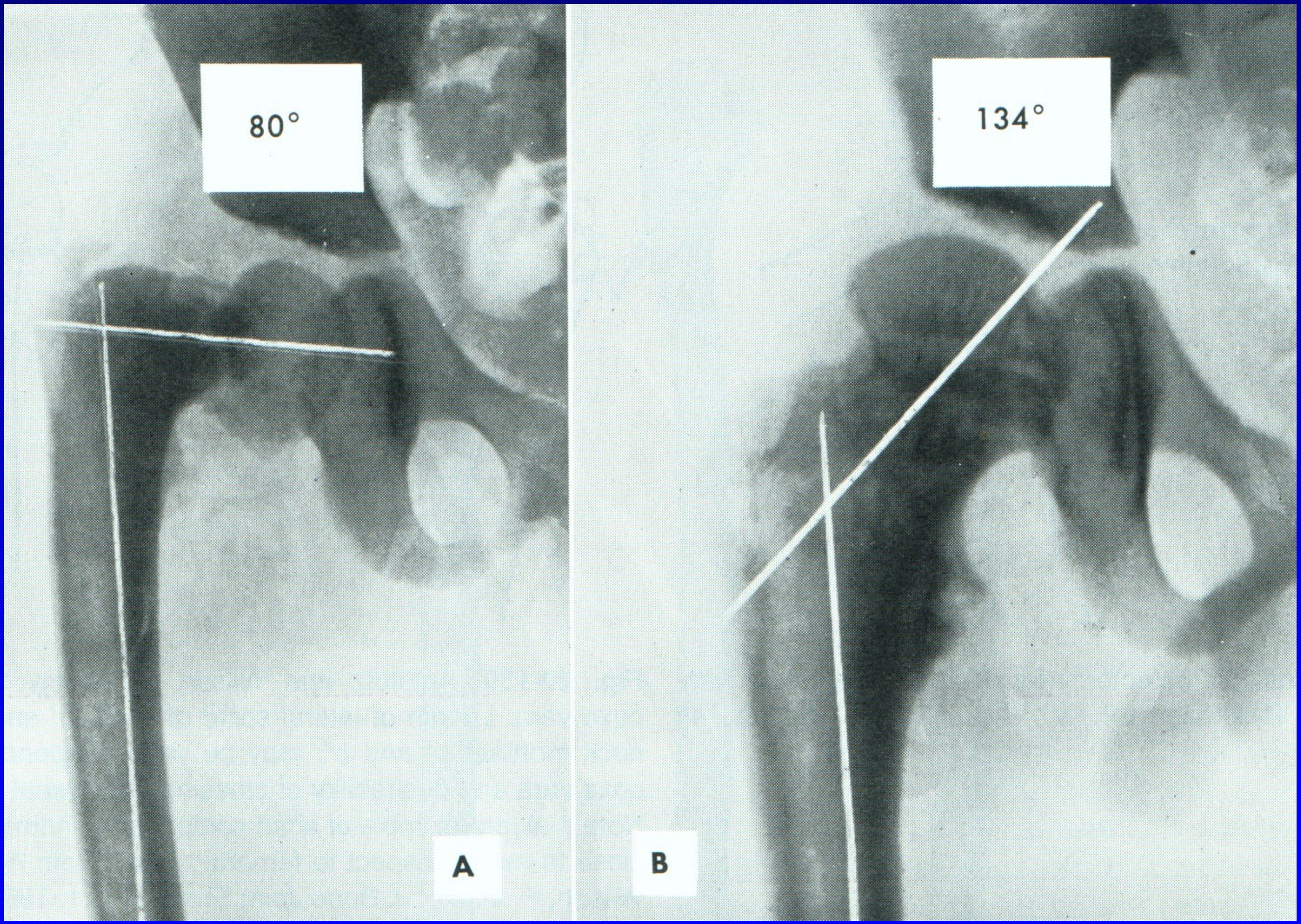
Obr. 12

Coxa vara congenita



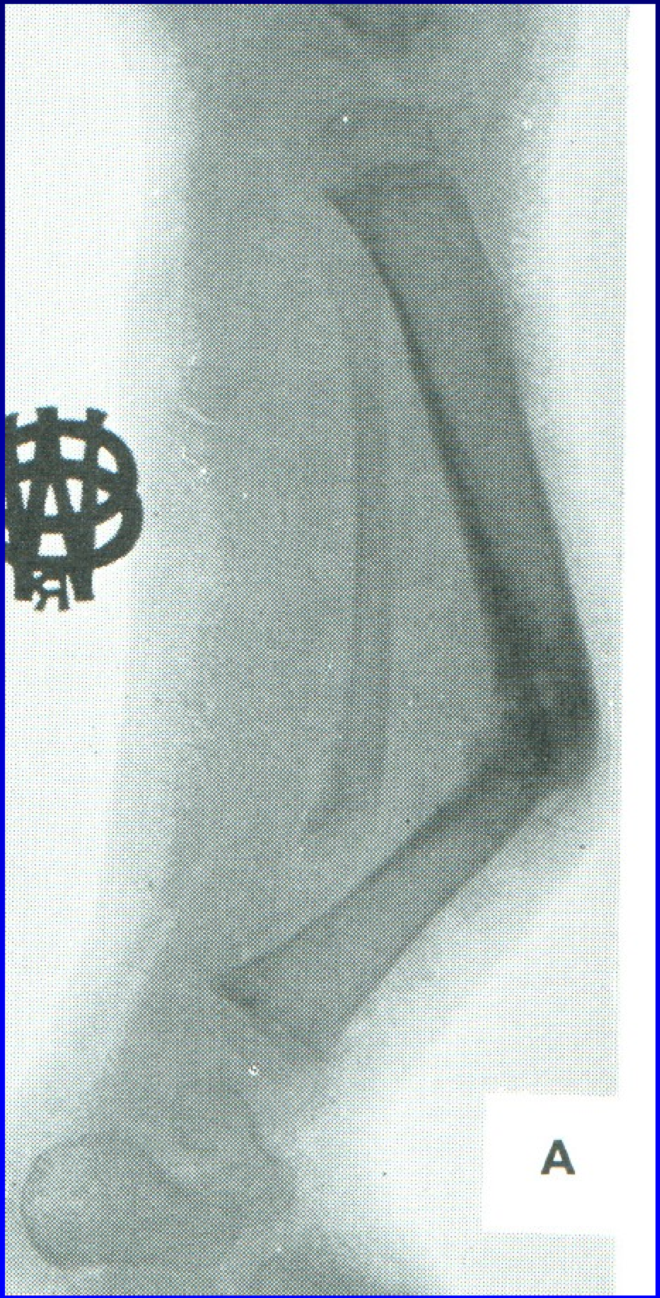
Obr. 13

Coxa vara congenita



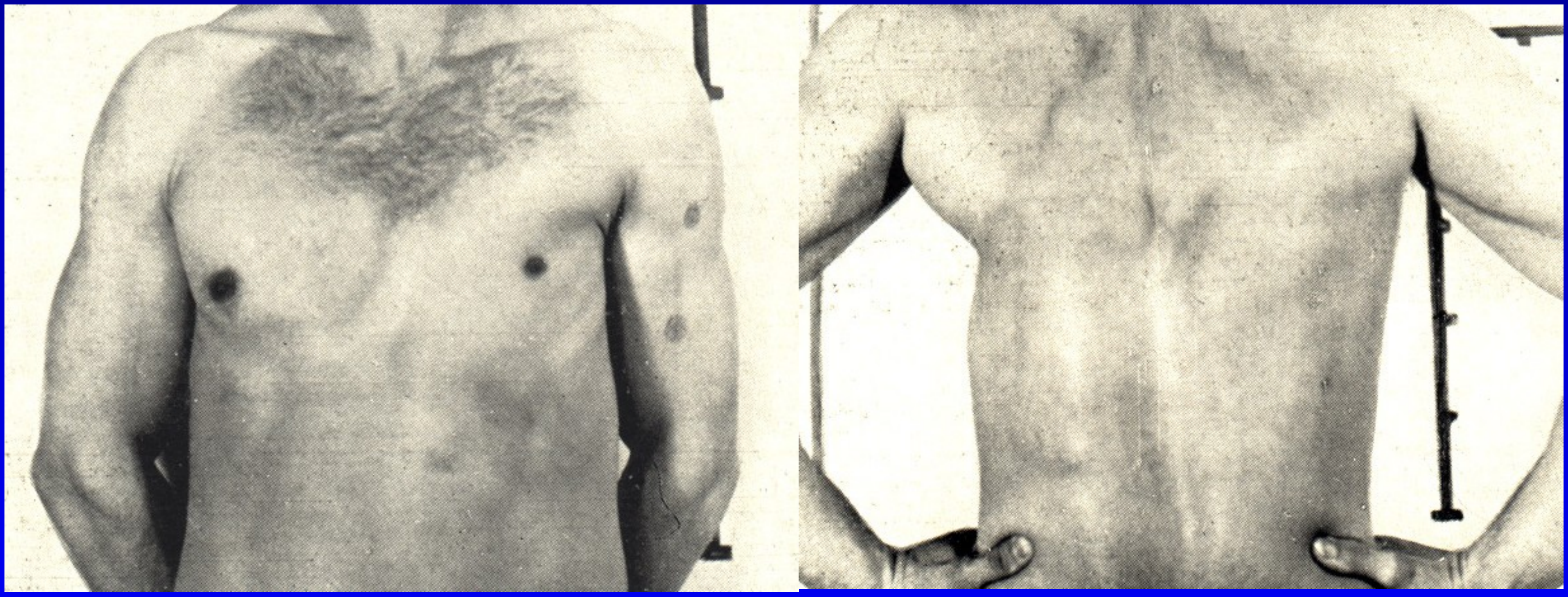
Obr. 14

Congenital nonunion of the tibia



Obr. 15

Aplasia of pectoral muscles

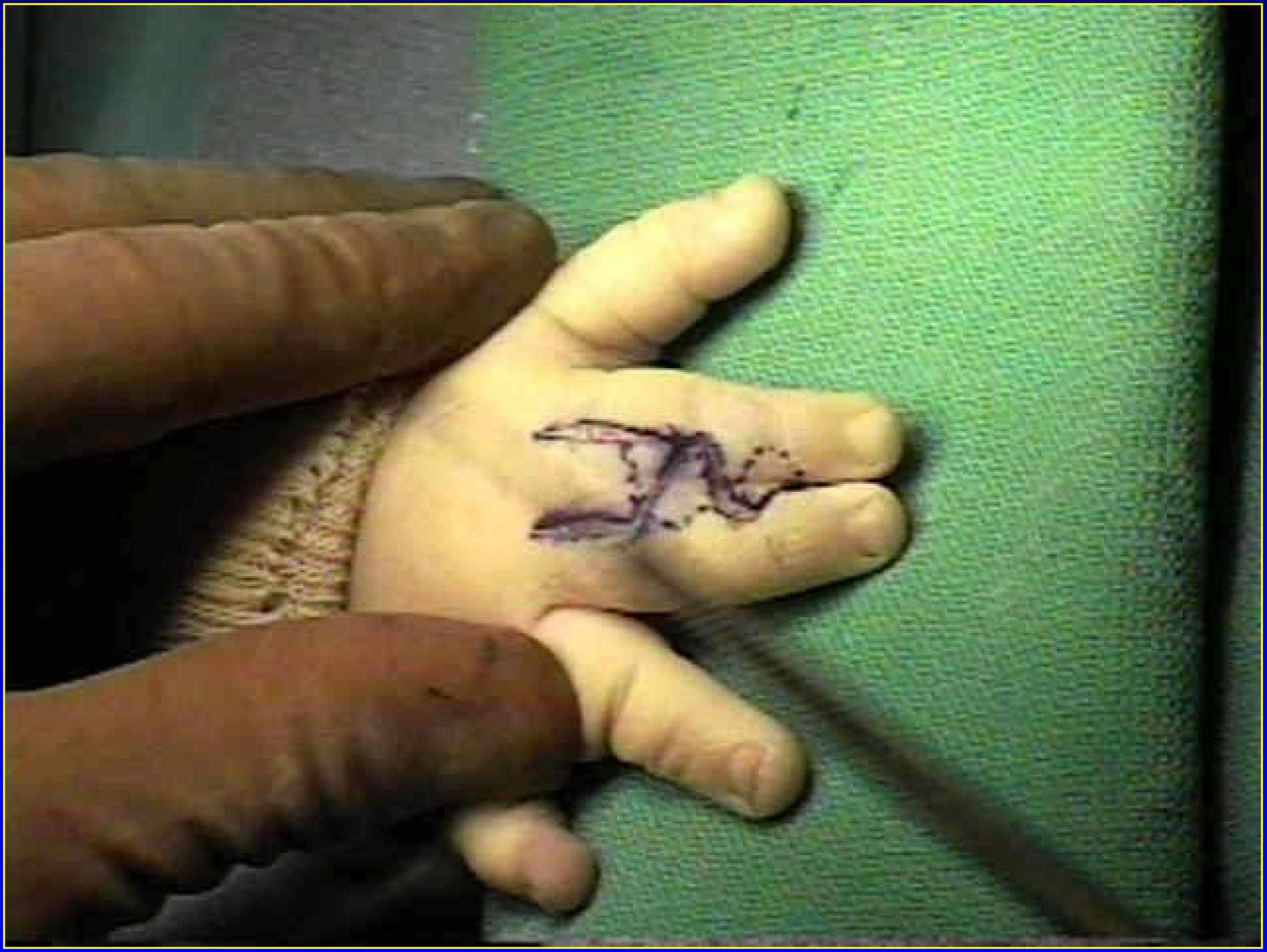


Obr. 16

2. Disorders of differentiation

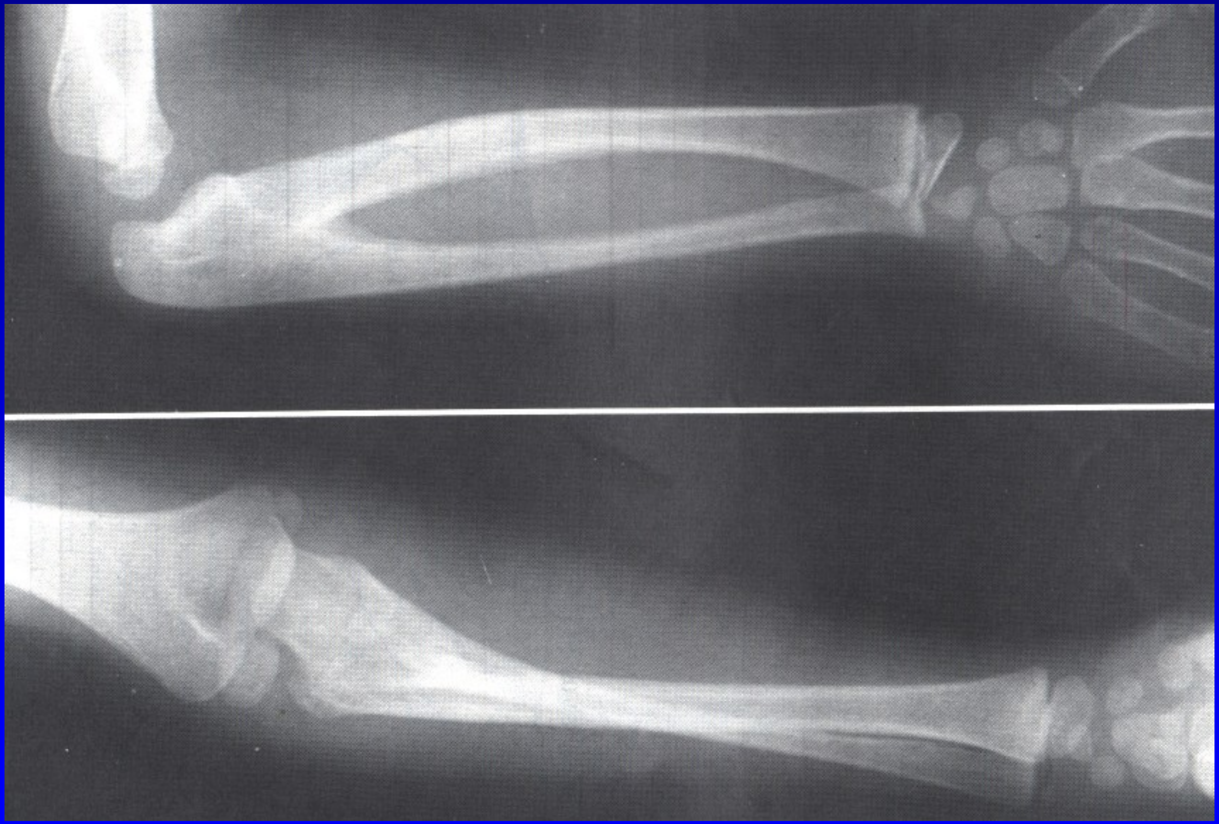
Syndactylia
Radioulnar synostosis
synostosis of carpal bones
Synostosis of vertebras
Tarsal coalition

Syndaktylia



Obr. 17

Radioulnar synostosis

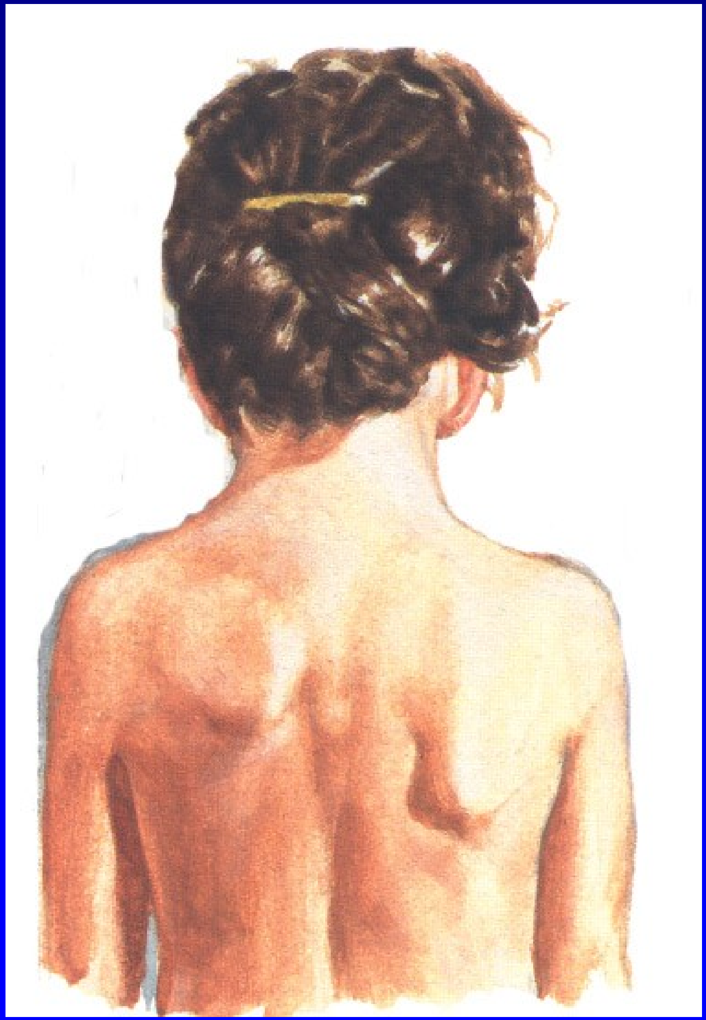


Obr. 18

Tarsal coalition

Obr. 19

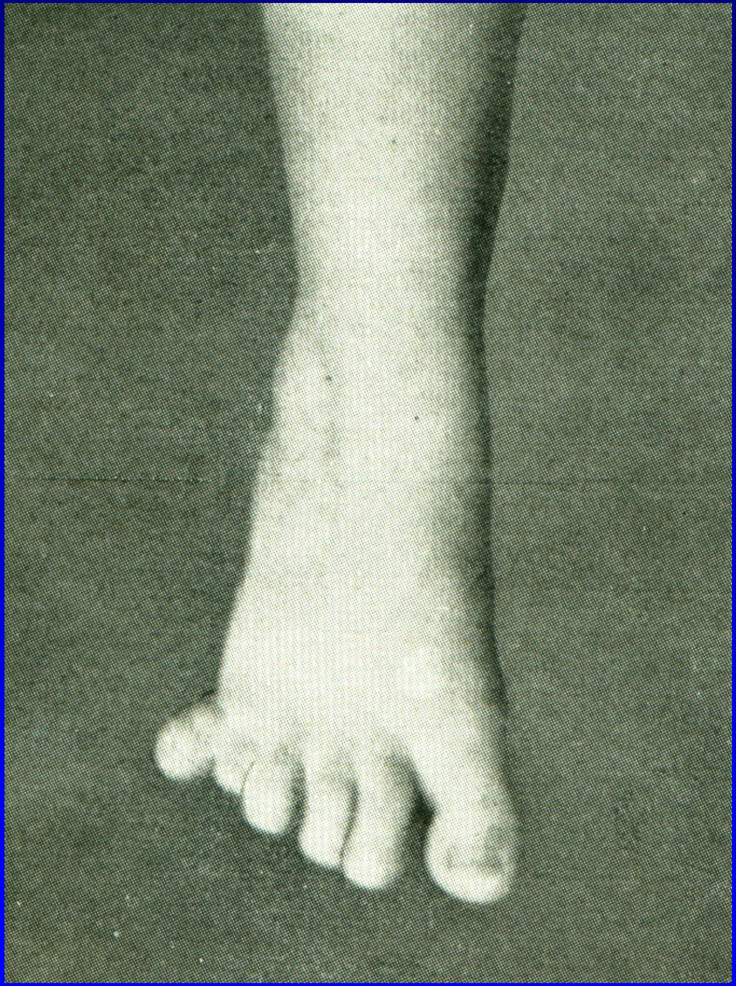
Sprengel´s deformity
- high scapula



Small scapula
Os omovertebrale
Fixed rotation of scapula
Limited movements

Obr. 20

3. Polydactylia



Obr. 21

Polydaktylia

Obr. 22

Polydaktylia

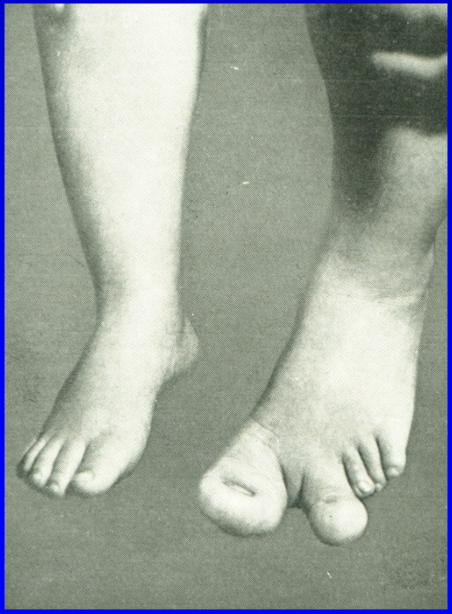
Obr. 23

4. Gigantisms

Growth hormon disorders

A- high stature (over 2 m)
 Gigantismus

B- hypertrophy of a part of body
 macrodactylia



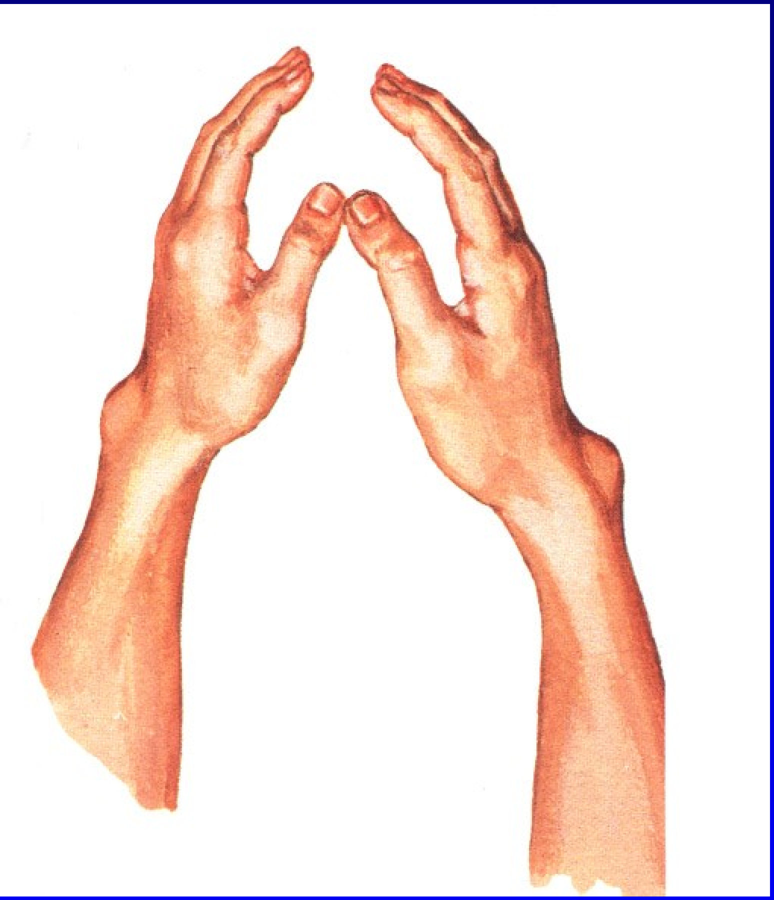
Makrodactylia

Obr. 25

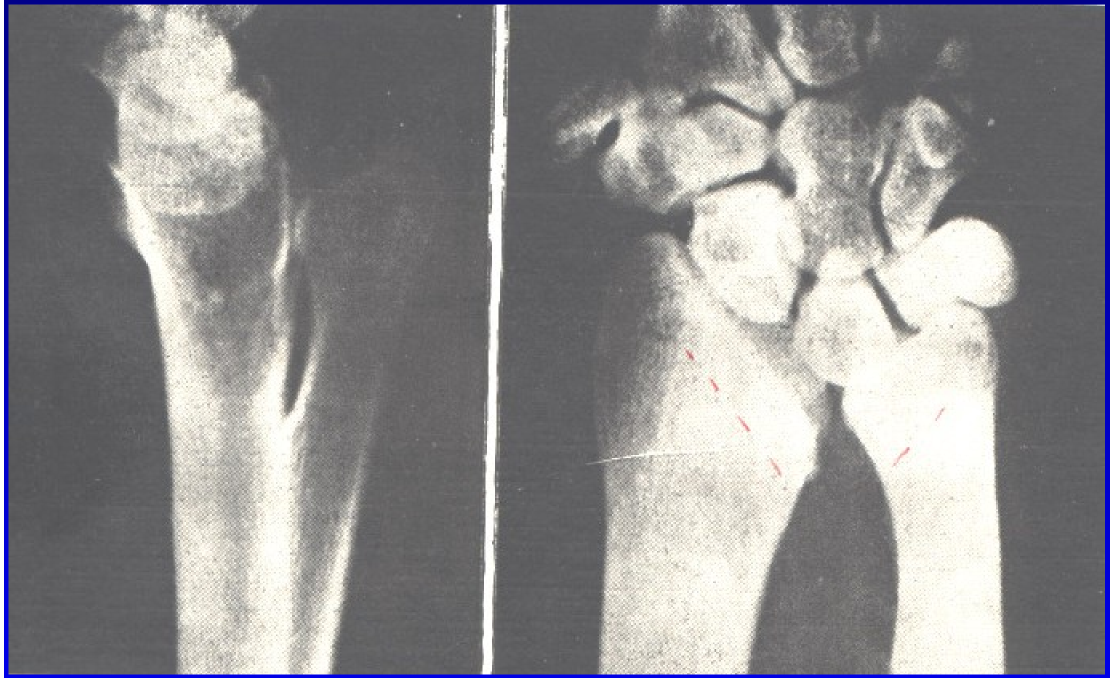
5. Hypoplasia

Brachydactylia
Brachyphalangia
Madelung deformity
Talipes equinovarus

Madelung deformity



Obr. 26



Obr. 27

Talipes equinovarus

Frequent disorder
Occurence 1/1000
Boys more often affected

1. Equinus of the ankle
2. Varus of the heel
3. Adduction of forefoot
4. Supination of the foot
5. Excavation of dorsum of the foot

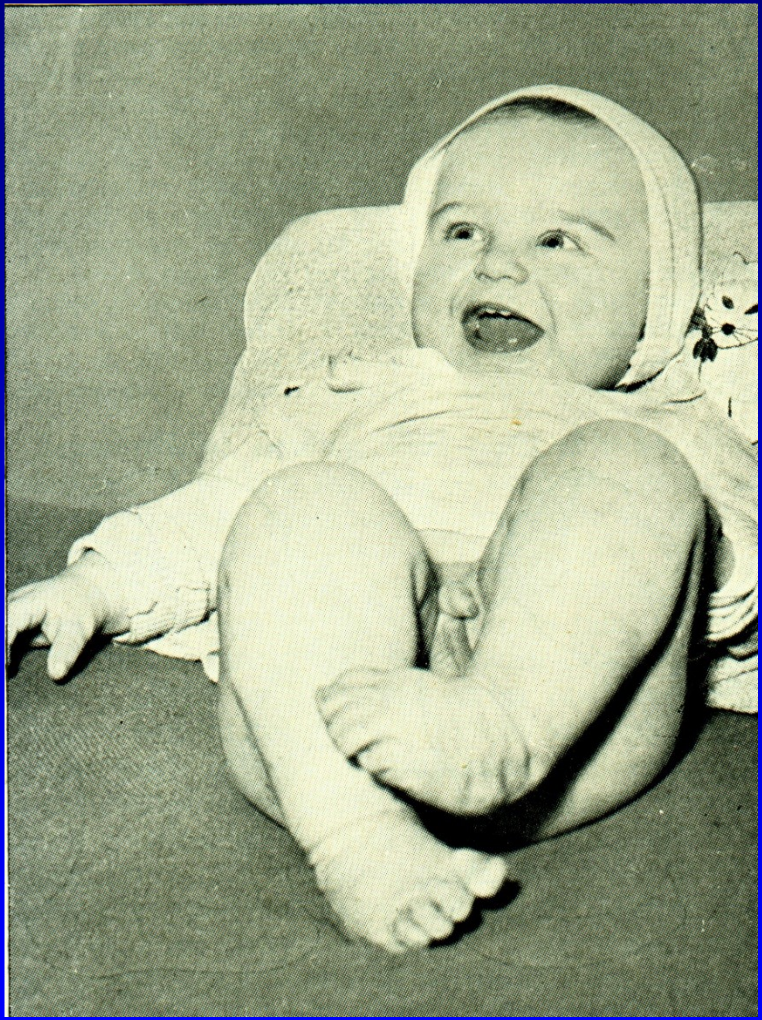
Obr. 28

Talipes equinovarus

Positional

Rigid

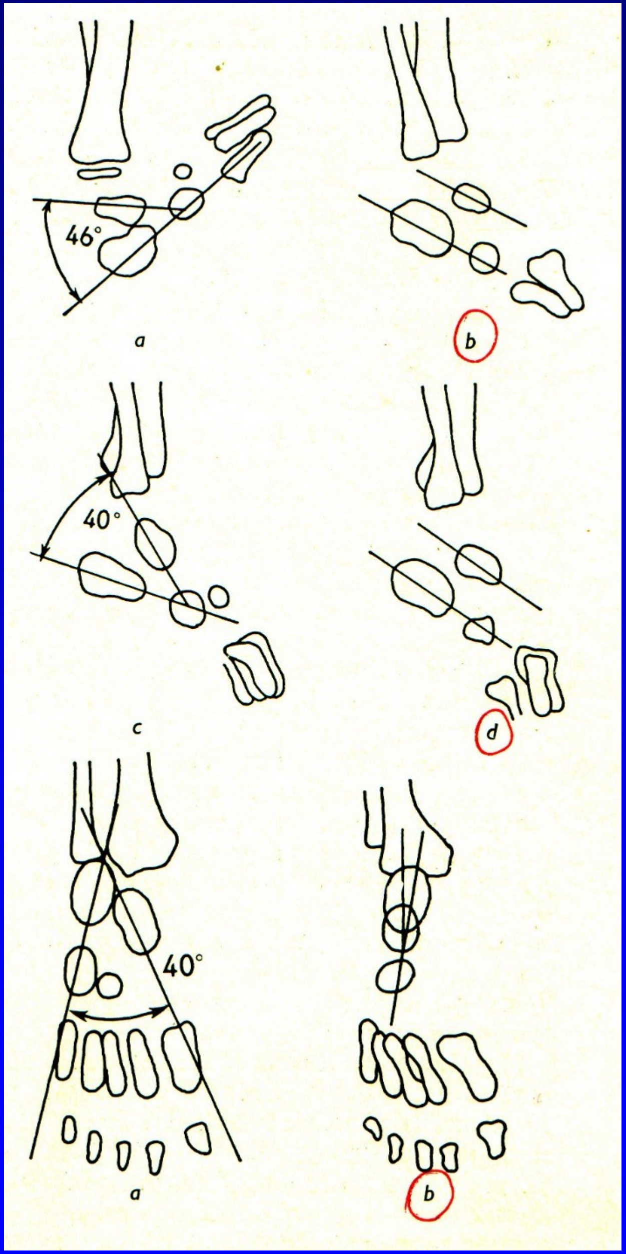
Rigid with other deformities



Obr. 28

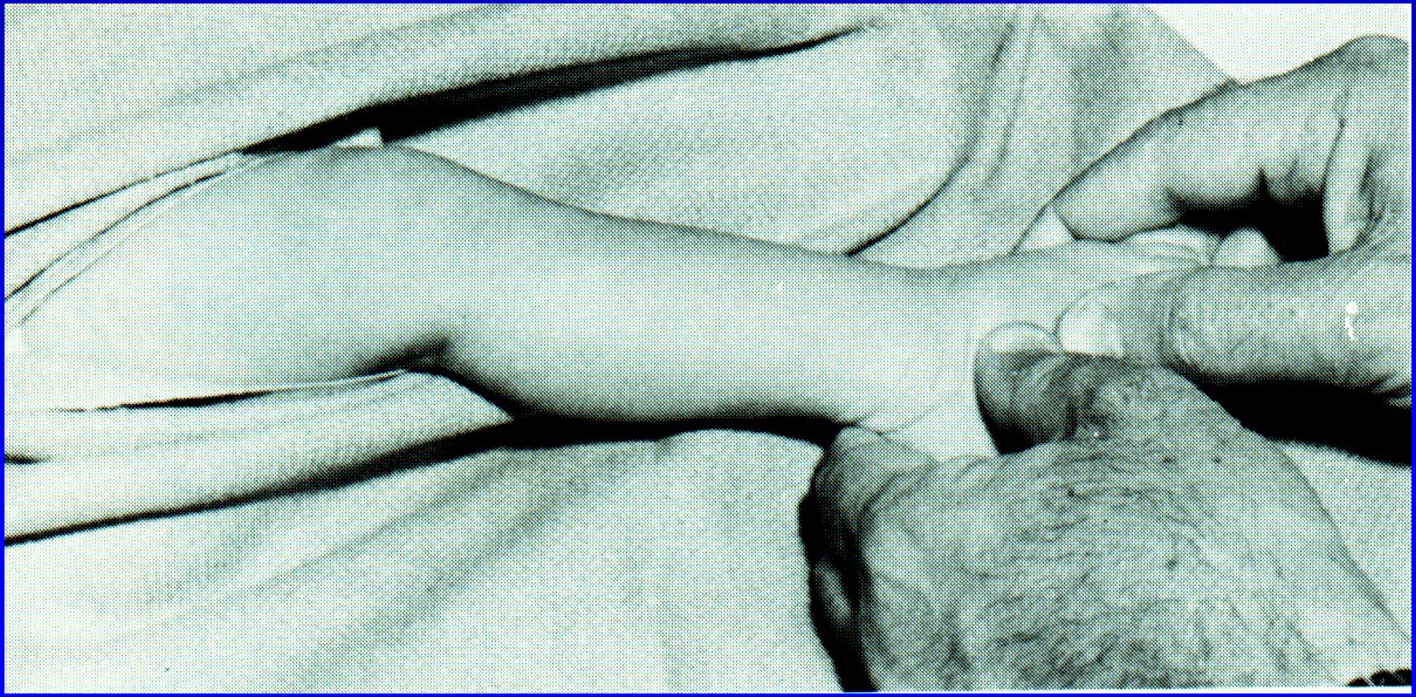
Pes equinovarus congenitus

X ray



Obr. 30

Management
Fysiotherapy – first 2 days



Obr. 31

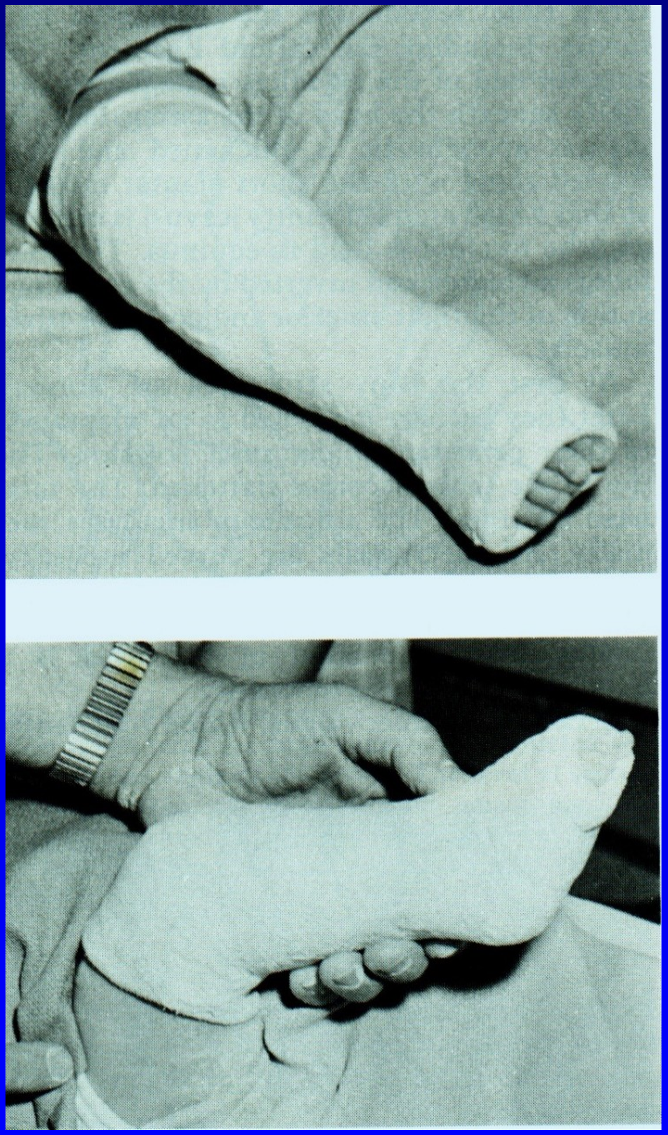
Splinting

First days after a birth

Change- twice weekly

10 weeks- 3 months

Correction of all deformities



Obr. 32

Ponseti method

Splinting 4 weeks +
correction of all deformities

Achilotomy + 6 weeks plaster

Denis- Brown splint

60 % succes
40 % surgery then necessary

Obr. 32

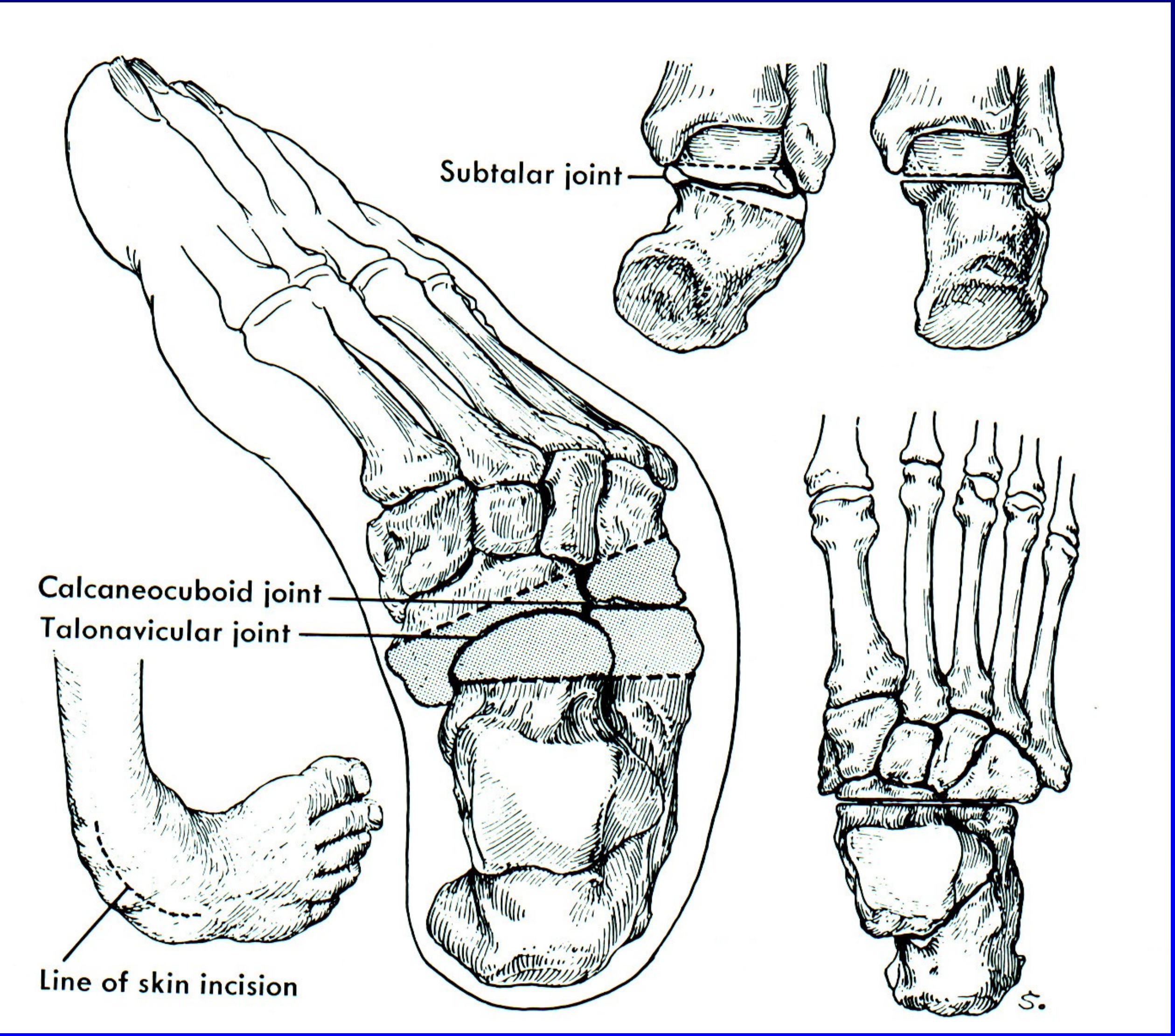
Surgery

Posterior capsulotomy +
lenghtening of Achillis tendon

Surgery- Turco, Mc Kay, Dwyer, Heymann, Bermann + Gartland.

Aftertreatment- plaster, correction splints

Triple arthrodesis



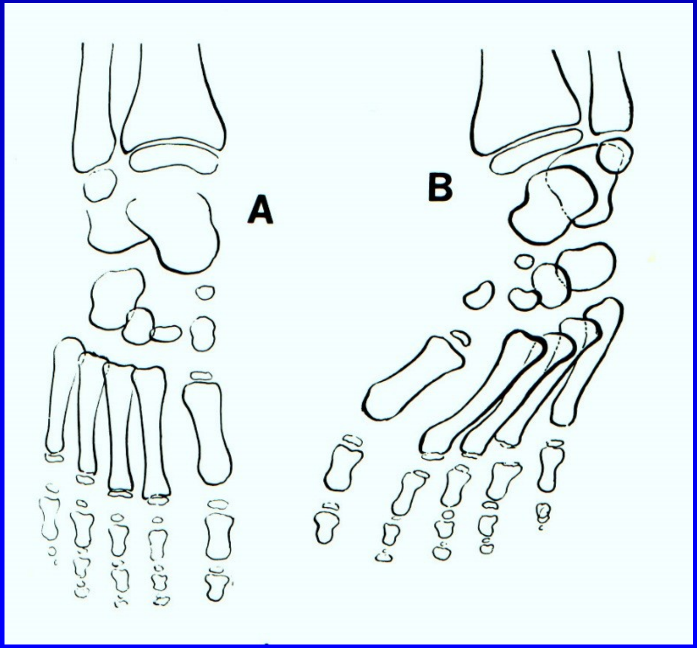
Obr. 34

Neglected pes equinovarus congenitus

Obr. 33

Metatarsus varus congenitus

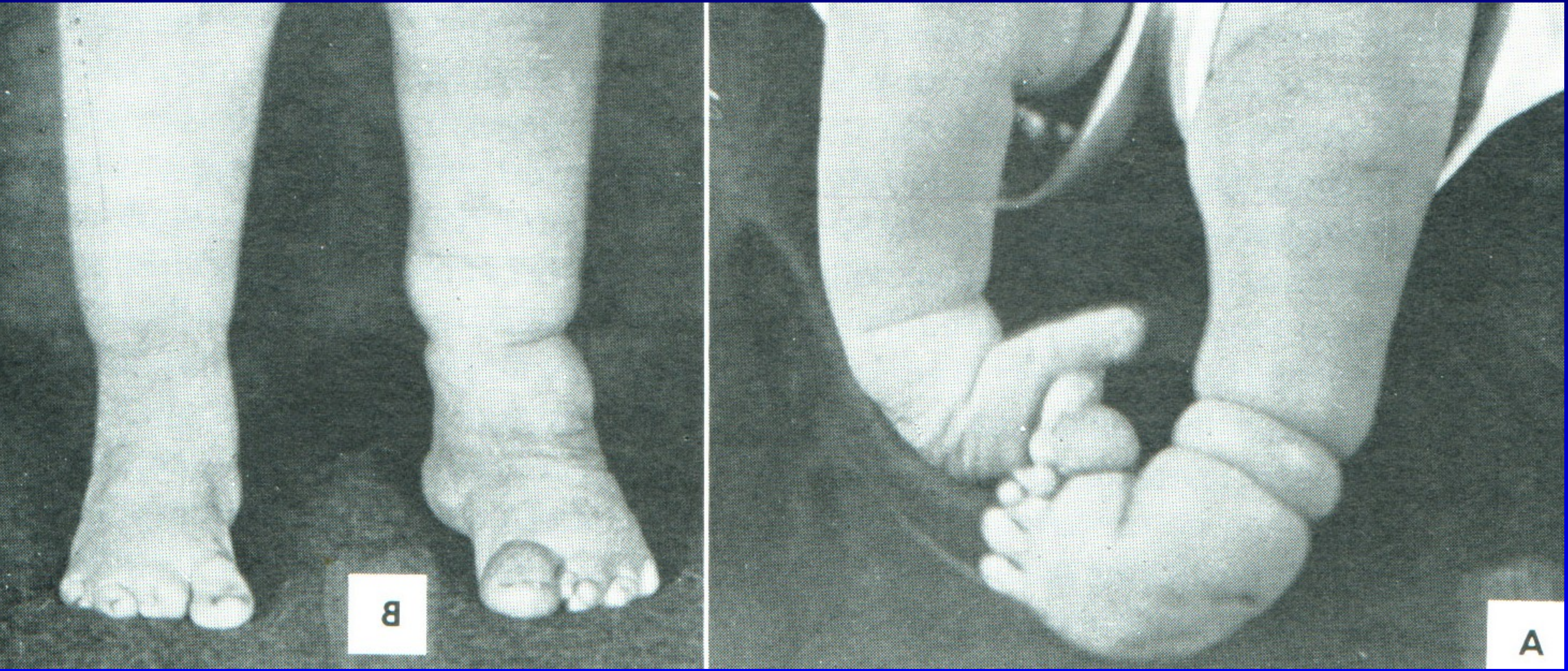
Mild form of equinovarus deformity
- adduction and supiation of the forefoot
Conservative and operative treatment



Obr. 35

Obr. 36

6. Congenital constrictions



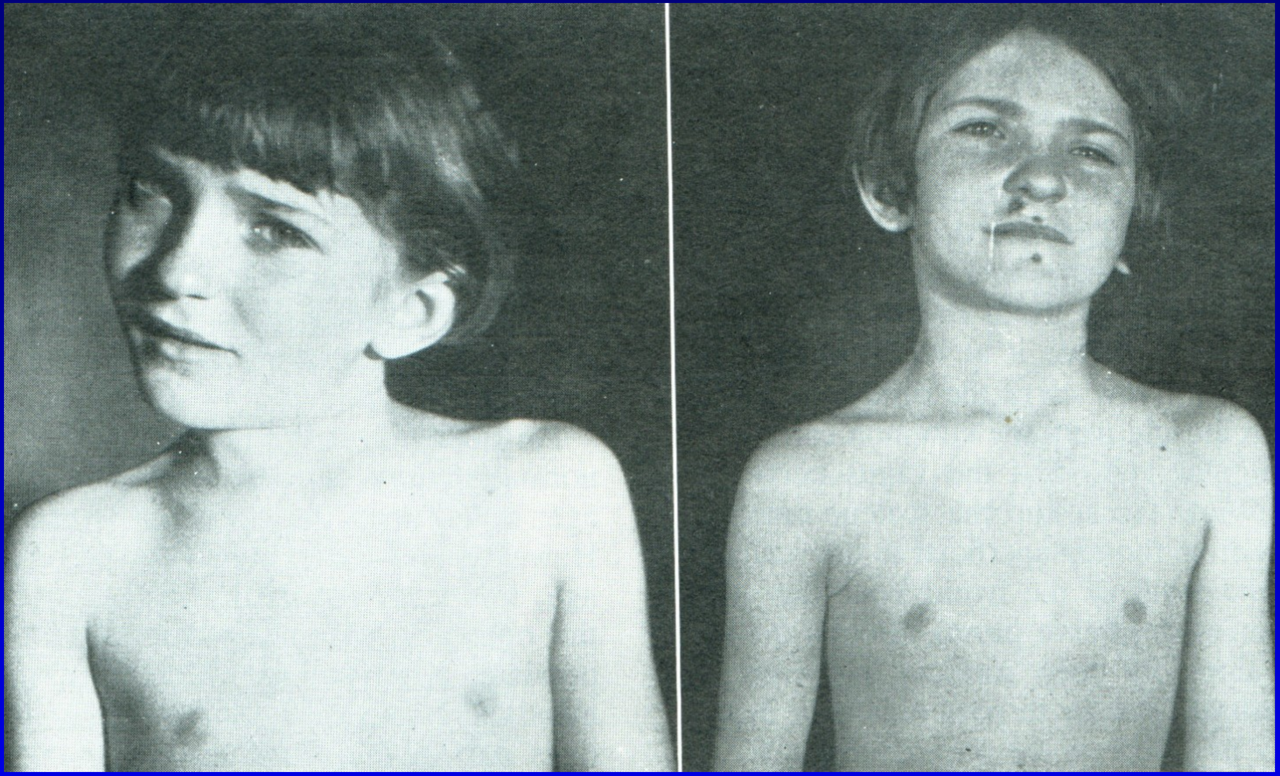
Obr. 37

Intrauterine development

Pterygium colli

Obr. 38

Wry neck- torticollis muscularis congenita



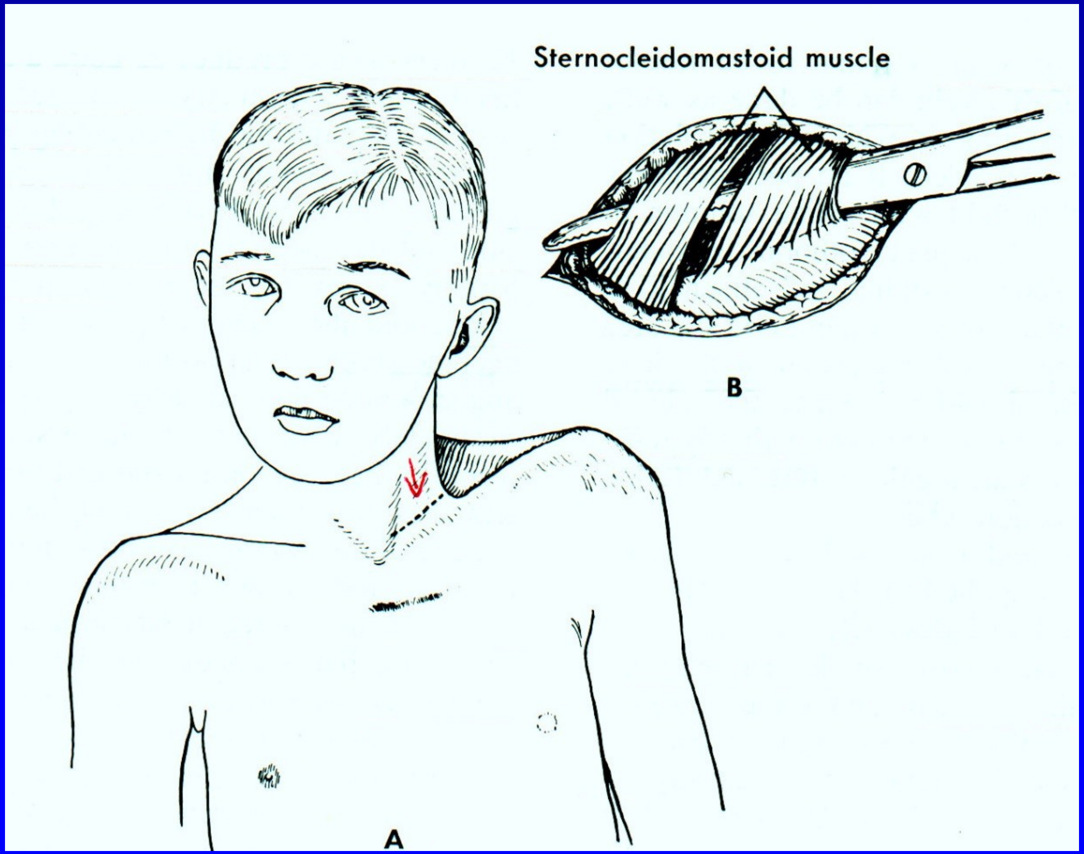
Obr. 39

Trauma of sternocleidomasteoideus muscle during delivery
Haematoma, tightening and fibrous shortening
The head – rotated to the healthy side
tilted to affected side
Asymetry of the face

Torticollis muscularis congenita

Positioning
Bandage

Surgery



General skeletal deformities

A. osseous

B. cartilage

C. mesenchymal

A. Osseous

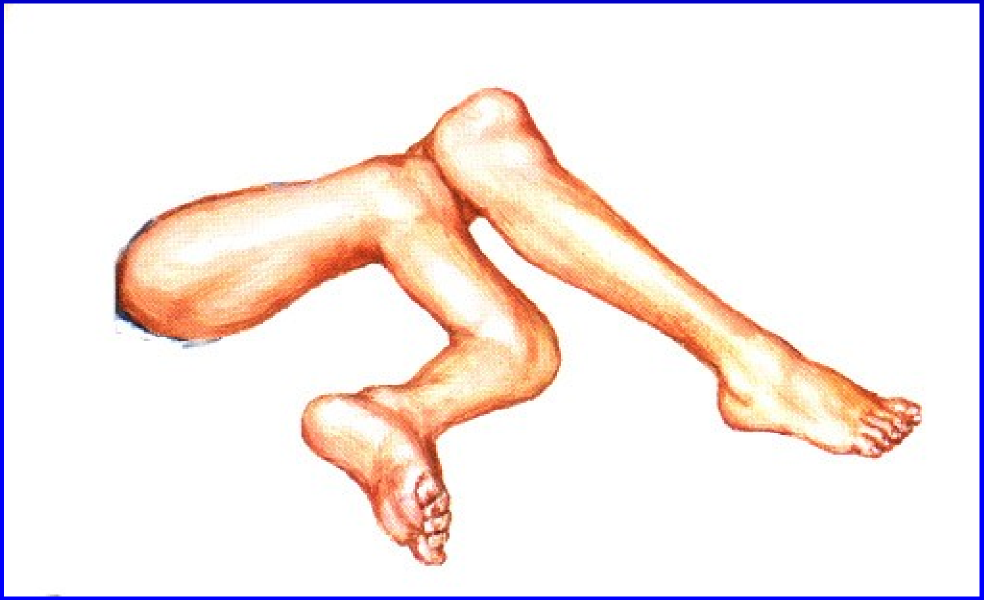
Osteogenesis imperfecta

Osteopetrosis

Osteogenesis imperfecta

I. – V. types
- letal (multiple fractures intrauterine)
- tarda (fractures, deformities, kyphosis,
  kyfoscoliosis, blue eyes
  deafness (otosclerosis)

Obr. 41



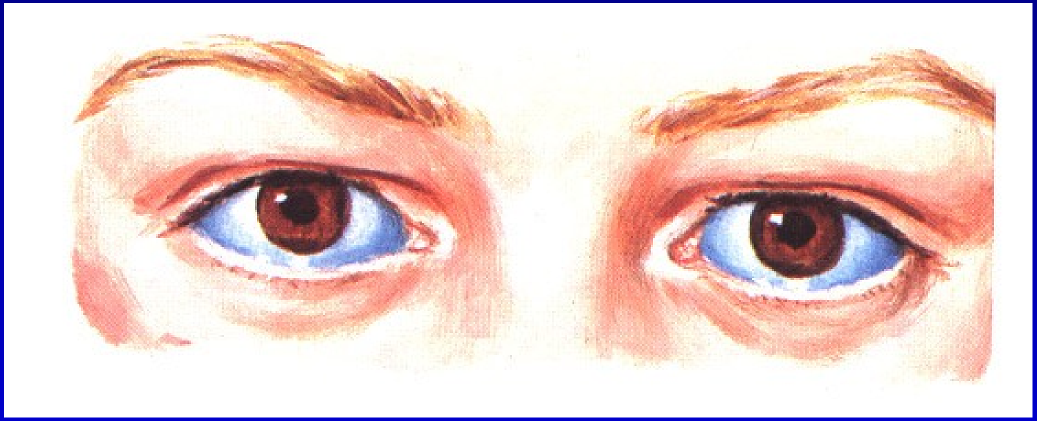
Obr. 42

Osteogenesis imperfecta

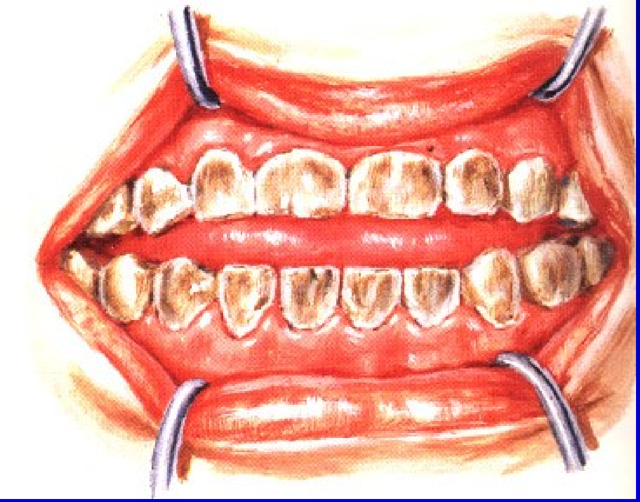
Obr. 43

Obr. 44

Osteogenesis imperfecta



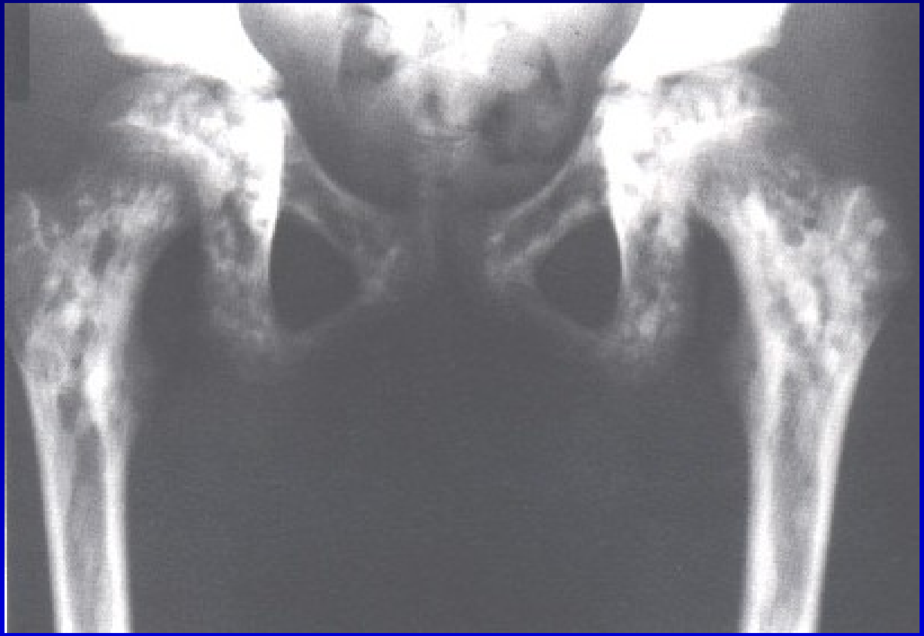
Obr. 45



Obr. 46

Osteopetrosis

Obr. 47



Obr. 48

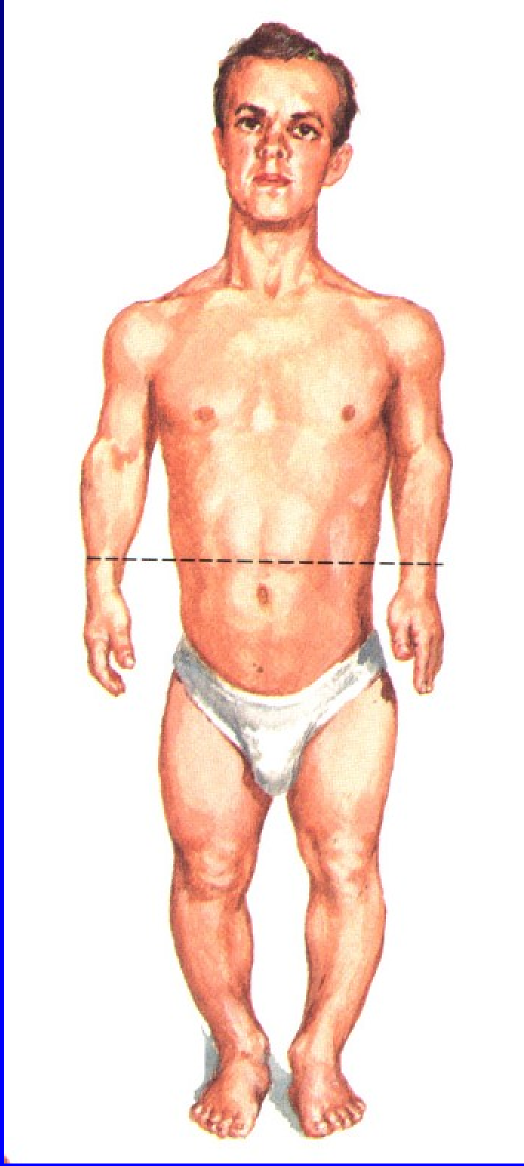
Osteopoikilosis

Osteopetrosis
Sclerotic and fragile bone

B. Cartilage

Achondroplasia
Multiple chondromatosis
Multiple epipsyseal dysplasia

Dwarfism- achondroplasia



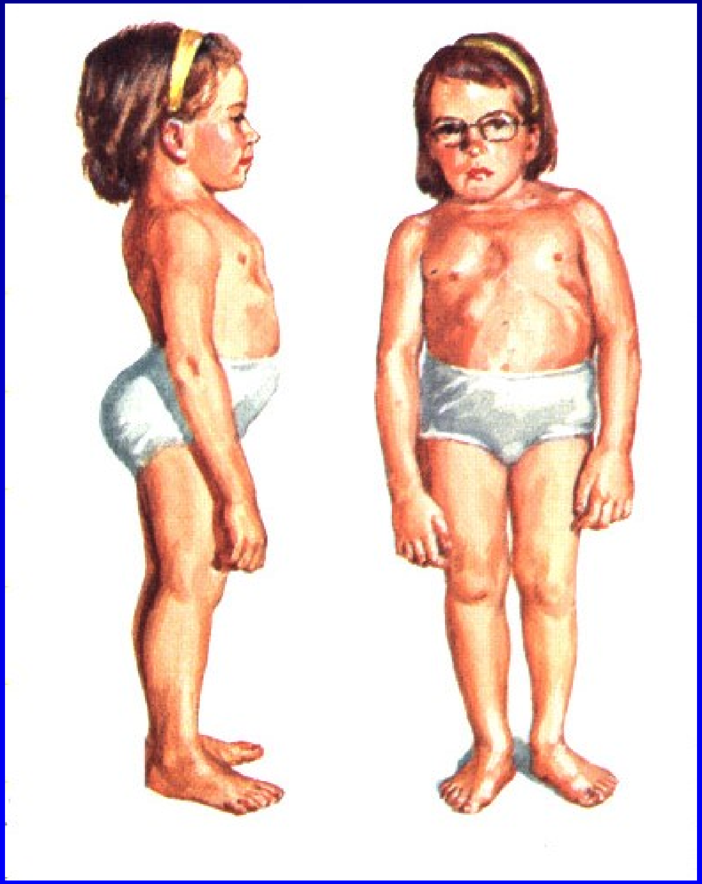
Dysproportional short stature
Long trunk, short extremities

Normal mental condition

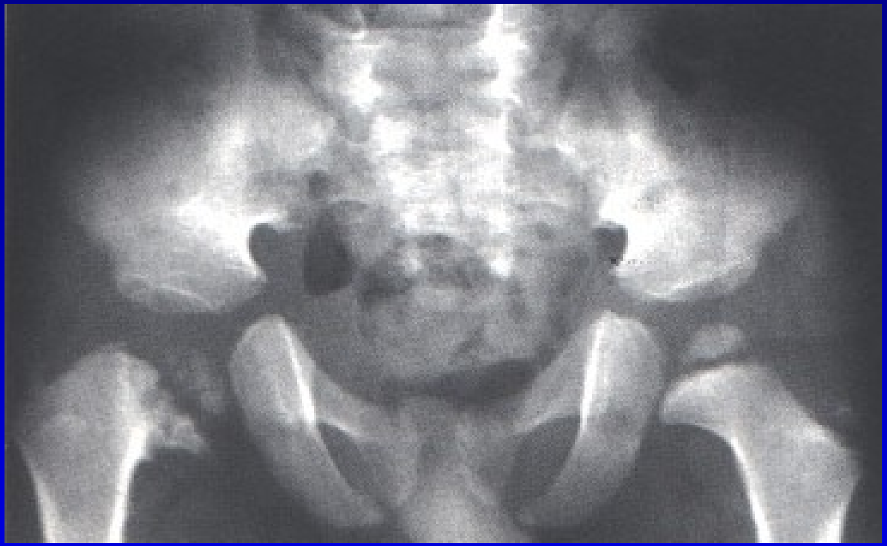
Low function of growth plates
with their premature closure

Obr. 49

Spondyloepifyseal dysplasia



Obr. 50



Obr. 51

Disorder of enchondral growth and ossifications
Deformity of epiphyseal region
Affections of joints, platyspondylia

Multiple osteochondromatosis

Obr. 52

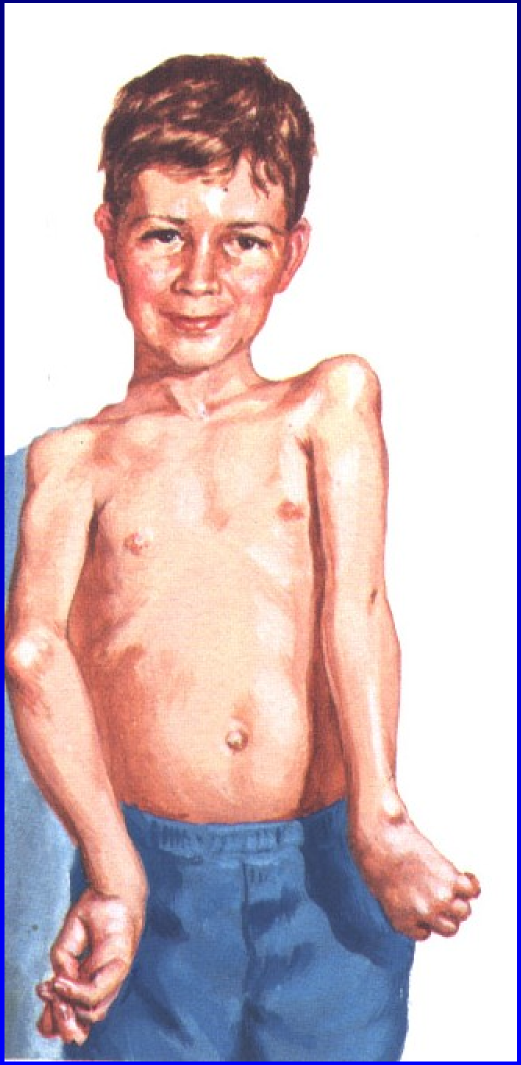
Obr. 53

Multiple osteochondroms

C. Mesenchymal

Arthrogryphosis congenita
Neurofibromatosis
Fibrous dysplasia
M. Ehlers-Danlos
Marfan syndrom
Mucopolysacharidosis

Arthrogyphosis congenita



Obr. 54

Change of muscles into fibrous and adiposis tissue

Stiff joints

In flexion
In extension

Normal mental condition

Neurofibromatosis

Obr. 55

Multiple neurofibroms

Caffe au lait patches

Large naevus

Hyperplasia

Hypoplasia

Neurofibromatosis

Obr. 56

Obr. 57

Kyfoskoliosis

Elefentiasis

Neurofibromatosis

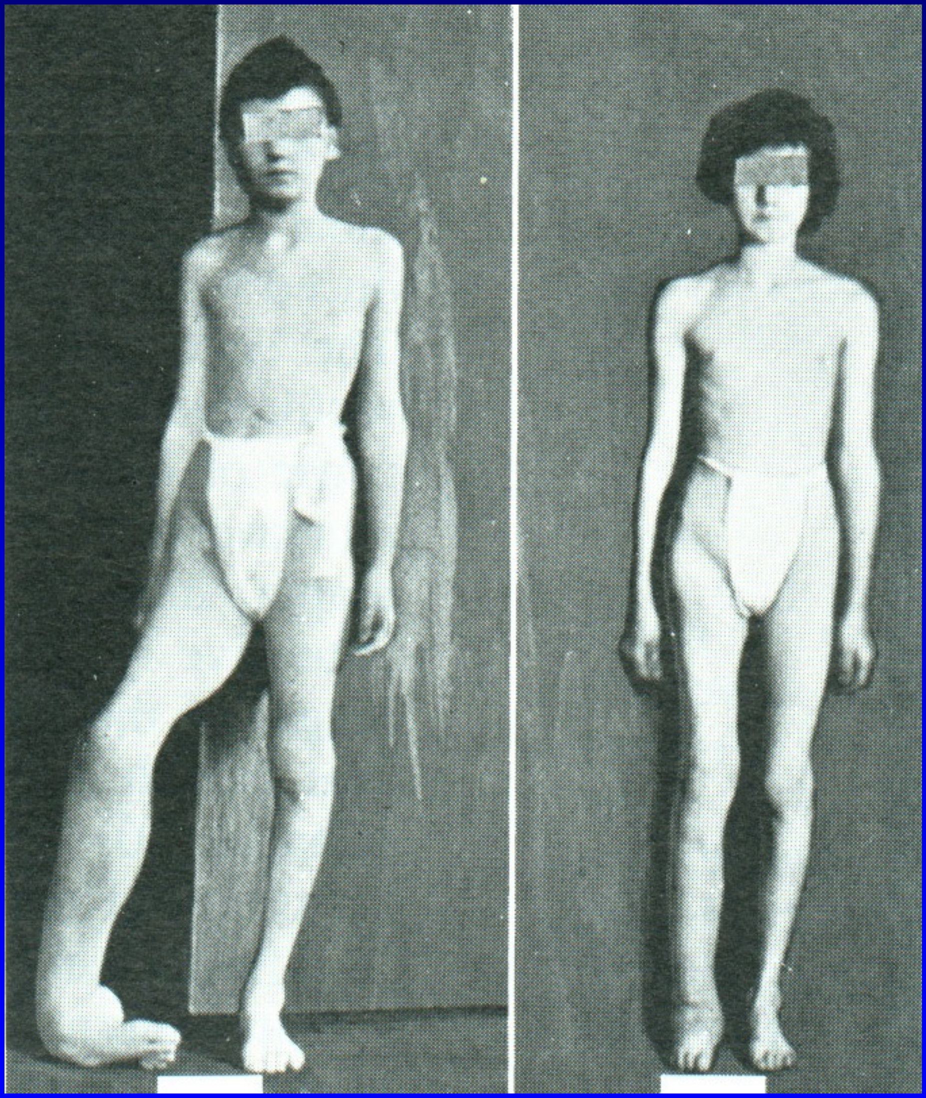
Oedema

Elefantiasis

Haemagiomas

Hyperplasia

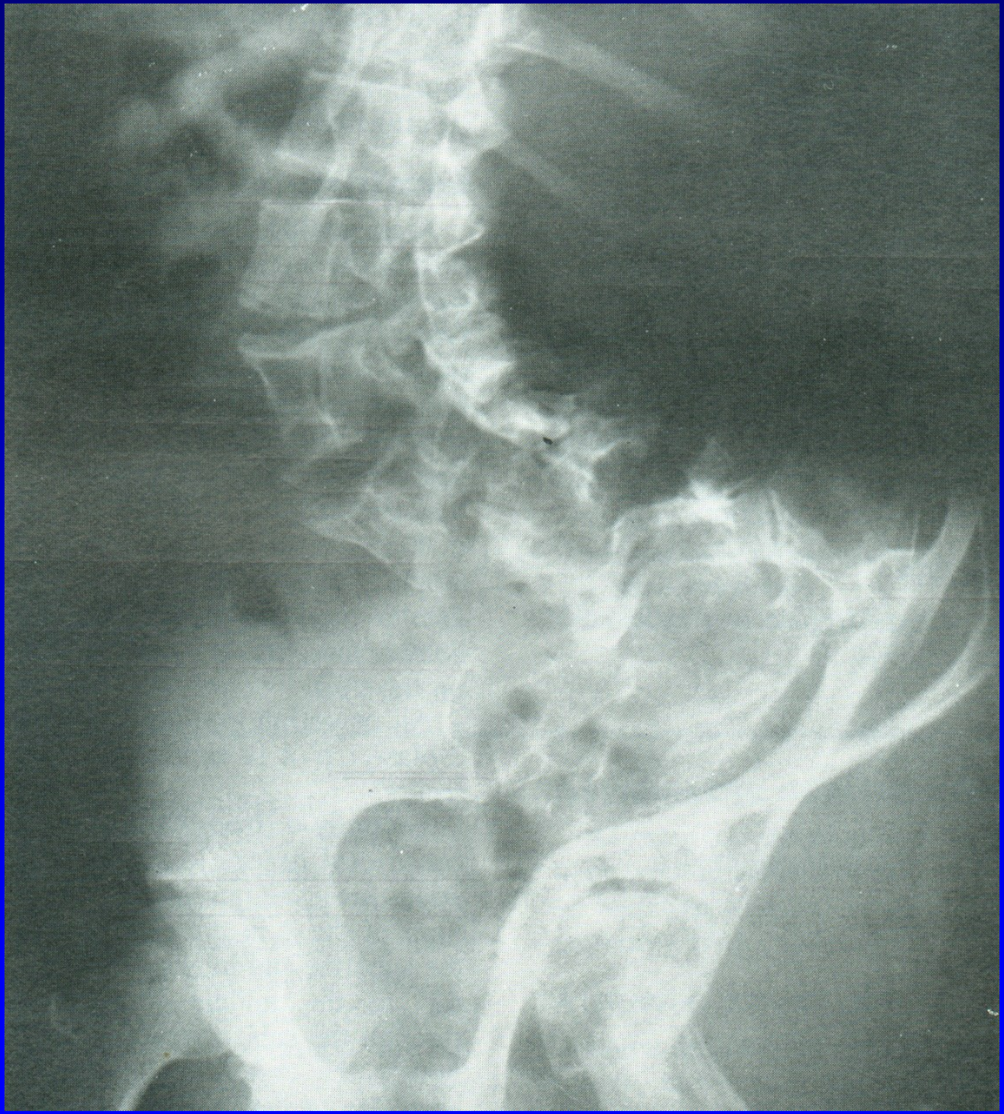
Hypoplasia



Obr. 58

Neurofibromatosis

Scoliosis
Deformity of the pelvis

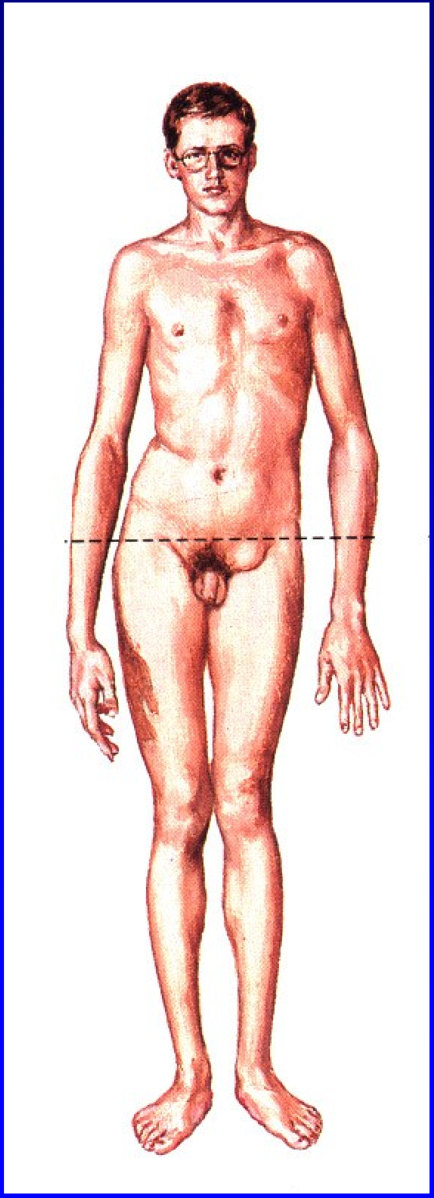


Obr. 59

Marfan syndrom

Tall stature
Long extremities
Long fingers
Scoliosis
Deformity of the strernum

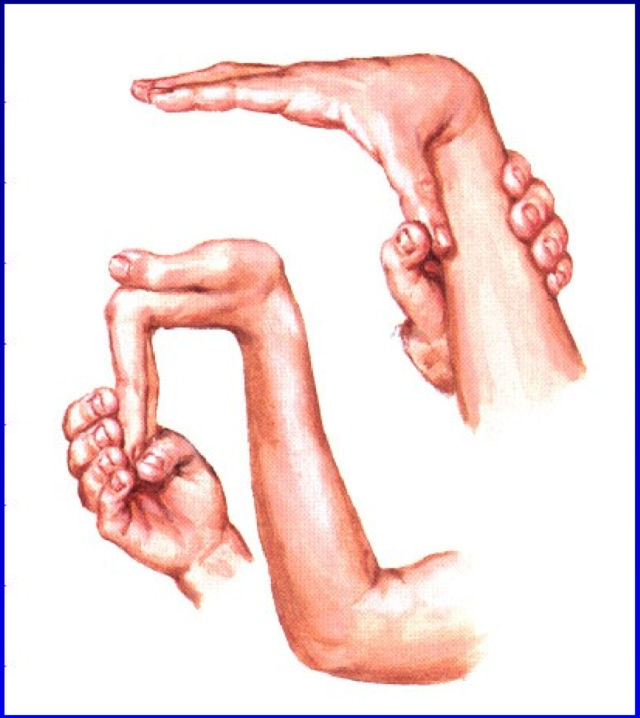
Aneurysma of the aorta
Hearth failure
Lens dystopy
Gothic patro



Obr. 60

Marfan syndrom

Obr. 61



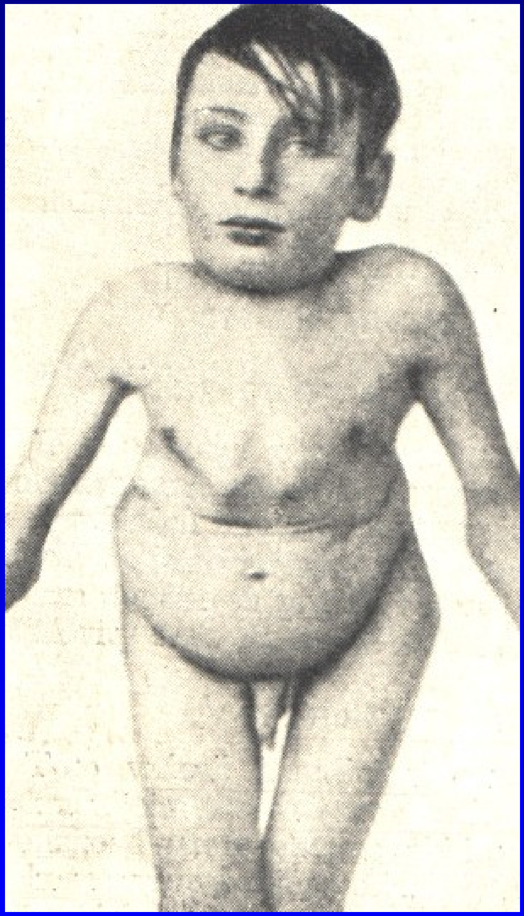
Obr. 62

Mesenchymal general laxity
genu recurvatum

Mucopolysacharidosis

Morquio- Brailsford syndrom
Hurler syndrom
Hunter syndrom

Morquio- Brailsford syndrom



Obr. 63

Platyspondylia
hyperkyphosis
Short stature
Short neck
Pectus carinatum
Weaknes of muscles
Hip deformities

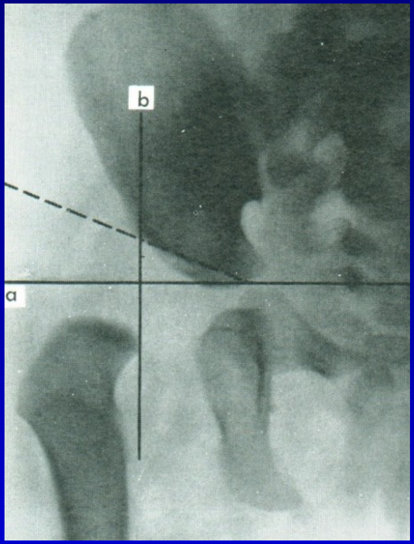
Developmental dysplasia of the hip

Occurence: 5 % of all children

Czech republic: 80-120
diclocations/ year

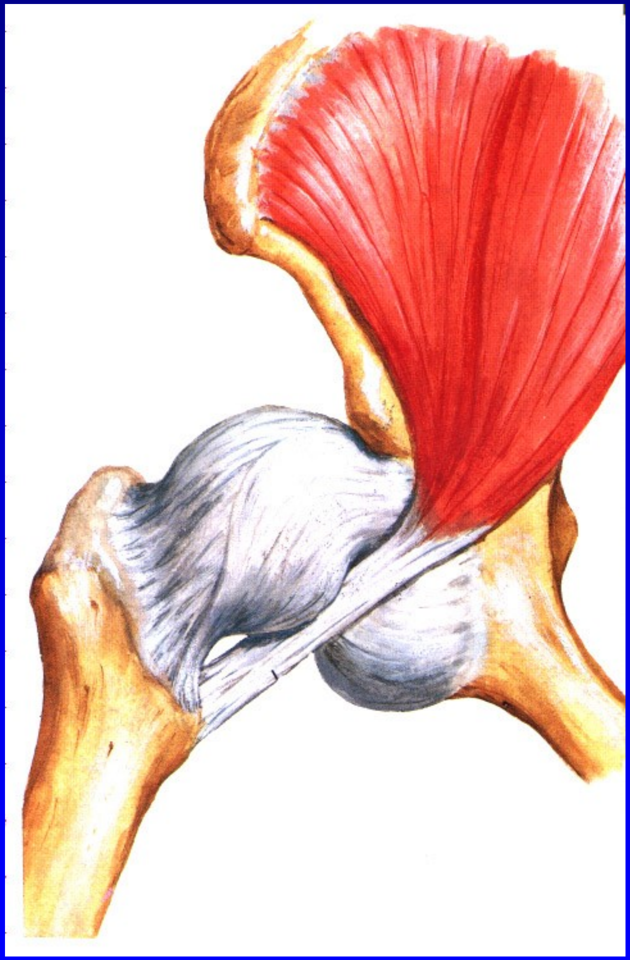
3-5 often girls

20 % of dysplastic O.A.



DDH

Perinatal and postnatal
Genes for acetabular dysplasia
Genes for joint capsule laxity
Increased laxity- relaxin
External forces
Birth- pelvis first



Obr. 64

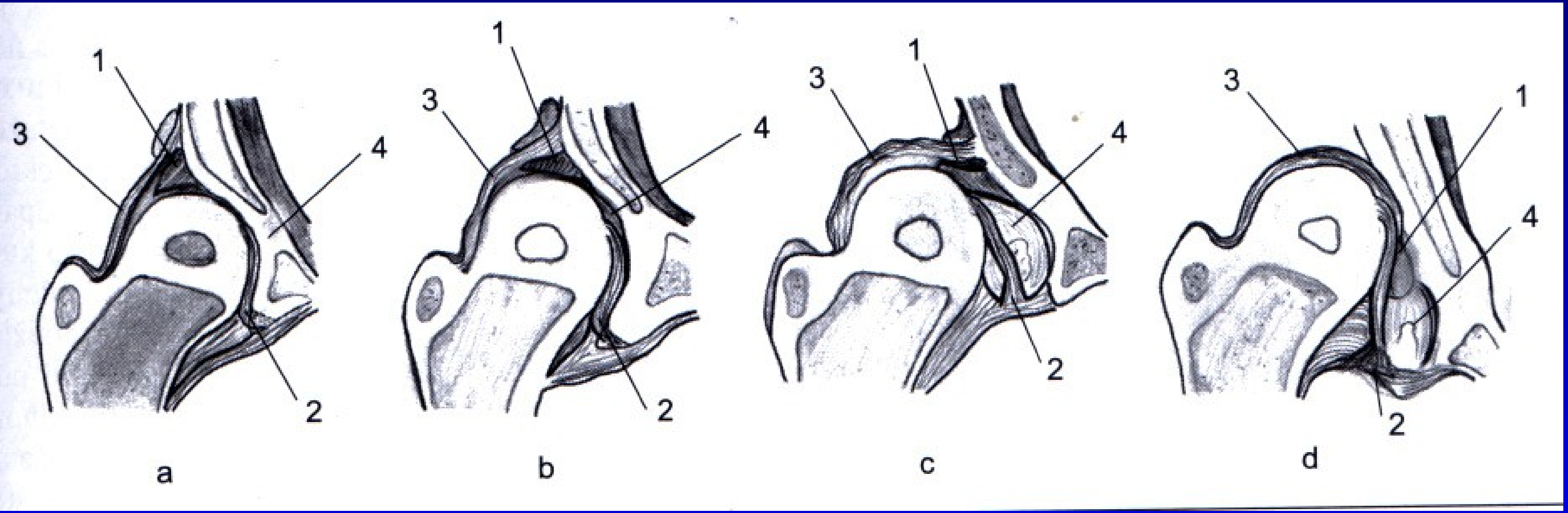
DDH

1. Dysplasia a- stable hip
   b- unstable hip
   steep osseous tectum
   late ossification of femoral head
   no decentration

2. Subluxation – head is subluxated

3. Dislocation- head is outside of the acetabulum

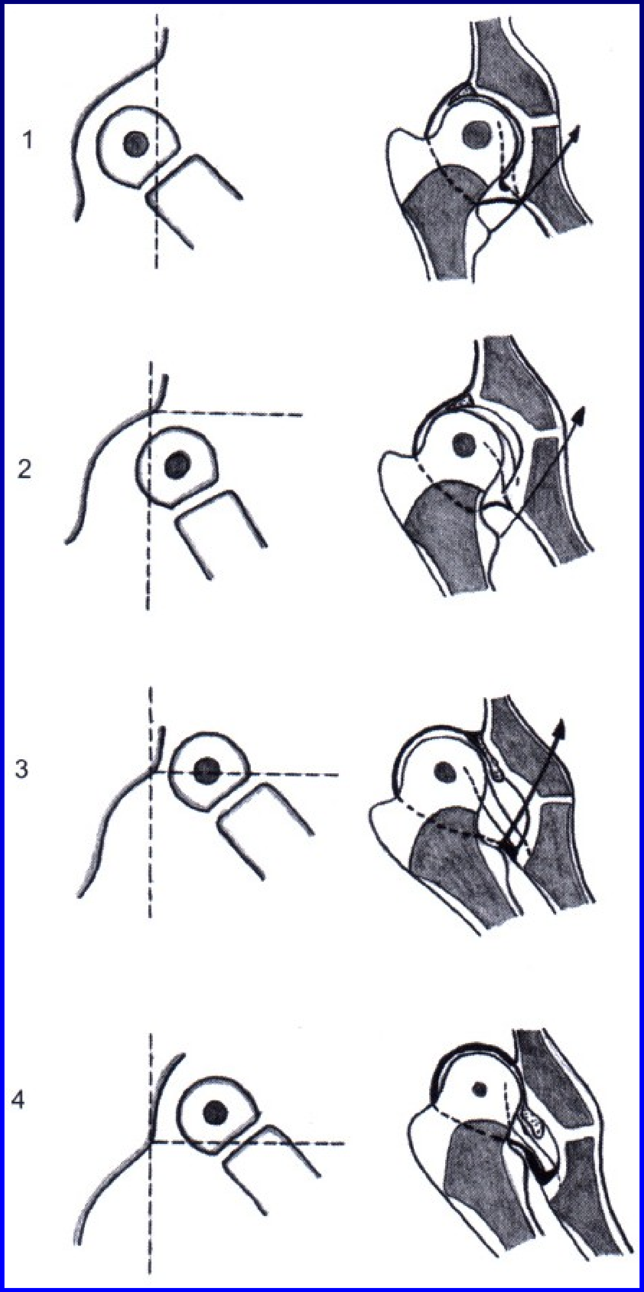
DDH



a dysplasia, stable hip
b dysplasia, unstable hip
c subluxation
d dislocation

AKH classification of DDH

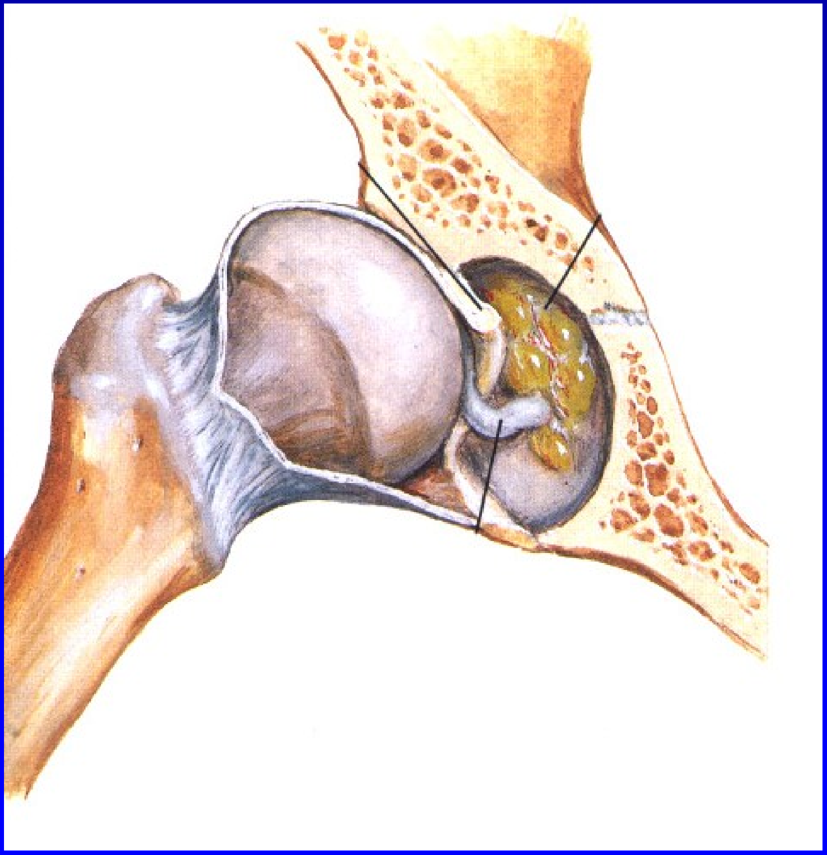
1 preluxation
2 unstable dysplasia
3 subluxation
4 dislocation



4.dislocation – head is outside of the acetabulum

luxatio marginalis
luxatio supracotyloidea
luxatio iliaca

Obr. 66



Symptoms in a newborn child- certain

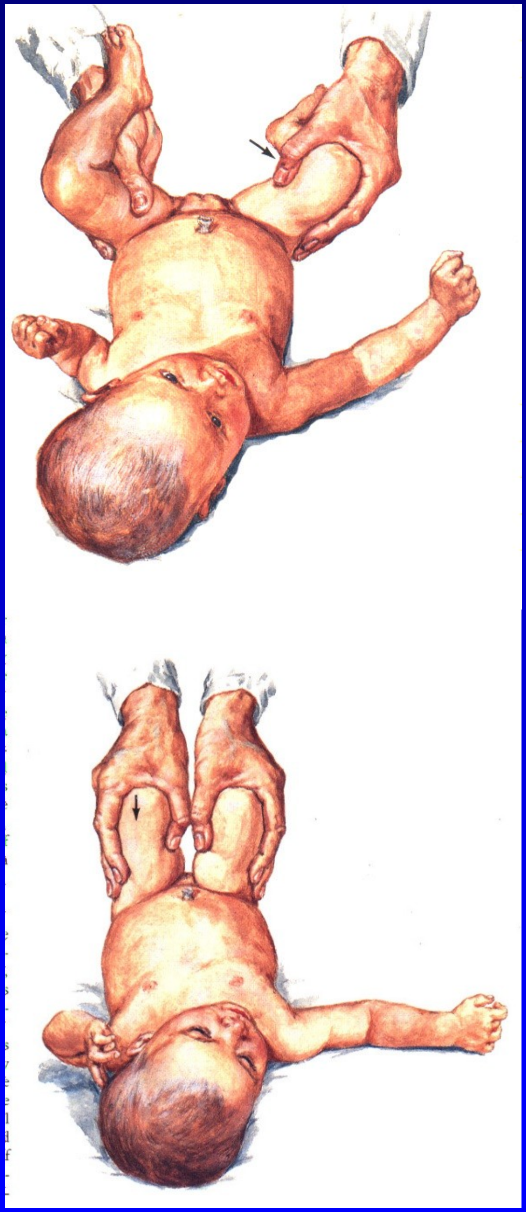
Ortolani abduction test (reduction)

Dislocation tests:

- Palmén test
- le Damany test
- Barlow test

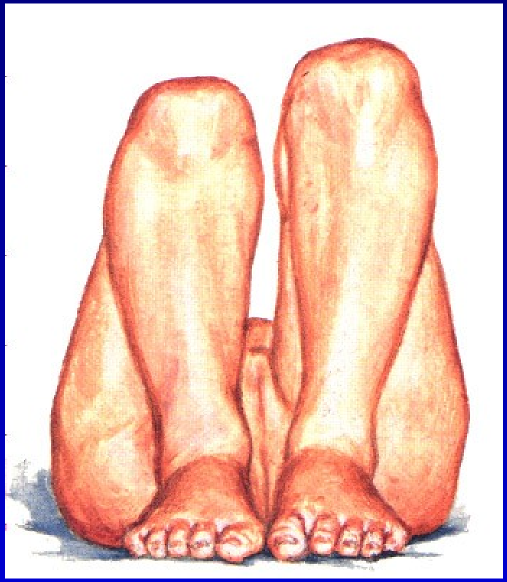
High position of greater trochynter

Femoral head is palpable
under abductors or in groin region

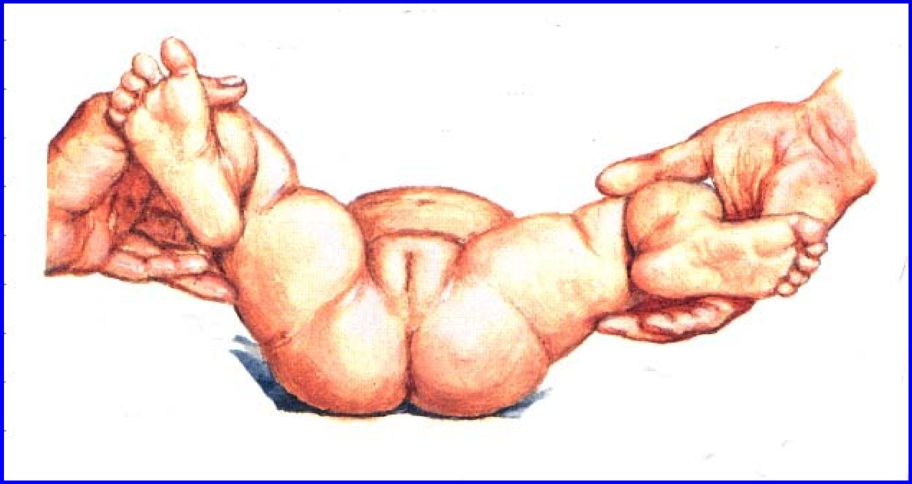


Obr. 67

Symptoms in a newborm child - suspision

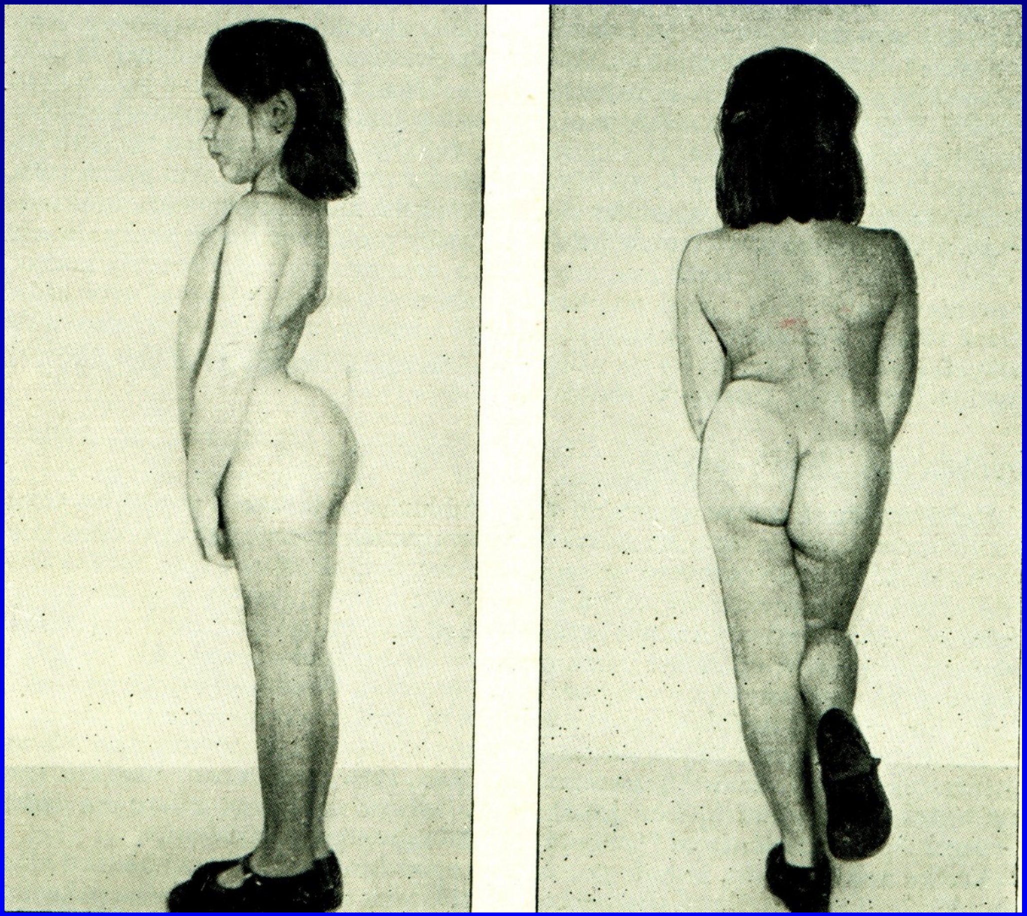


Shortening of the extremity
Limited abduction
Asymetry of skin grooves
Increased tonus of adductors



Symptoms in a older child

Limping
Shortening of the extremity
Trendelenburg sign
Lumbar hyperlordosis
Rocking child in
bilateral cases

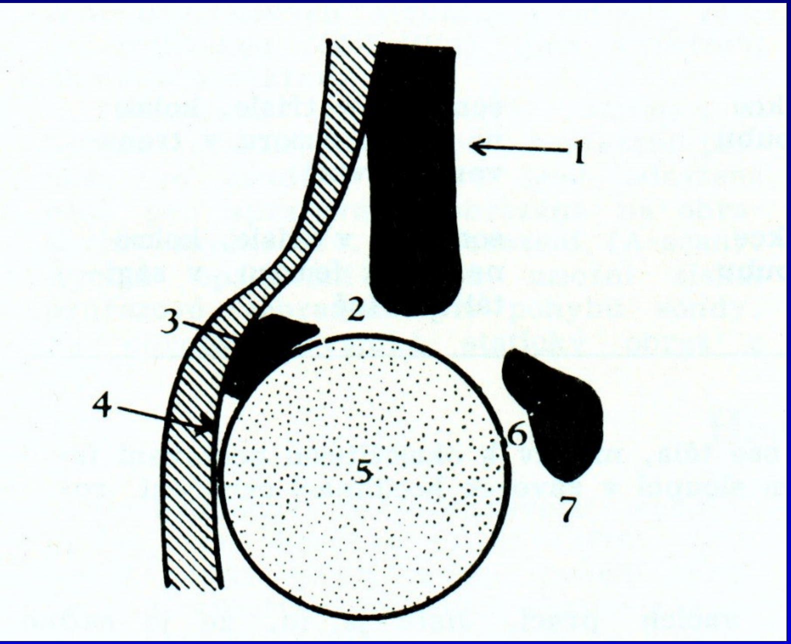


Obr. 70 Trendeleburg sign

Ultrasonography

Graf classification

1a
1b
II a
II b
II c
II d
III a
III b
IV



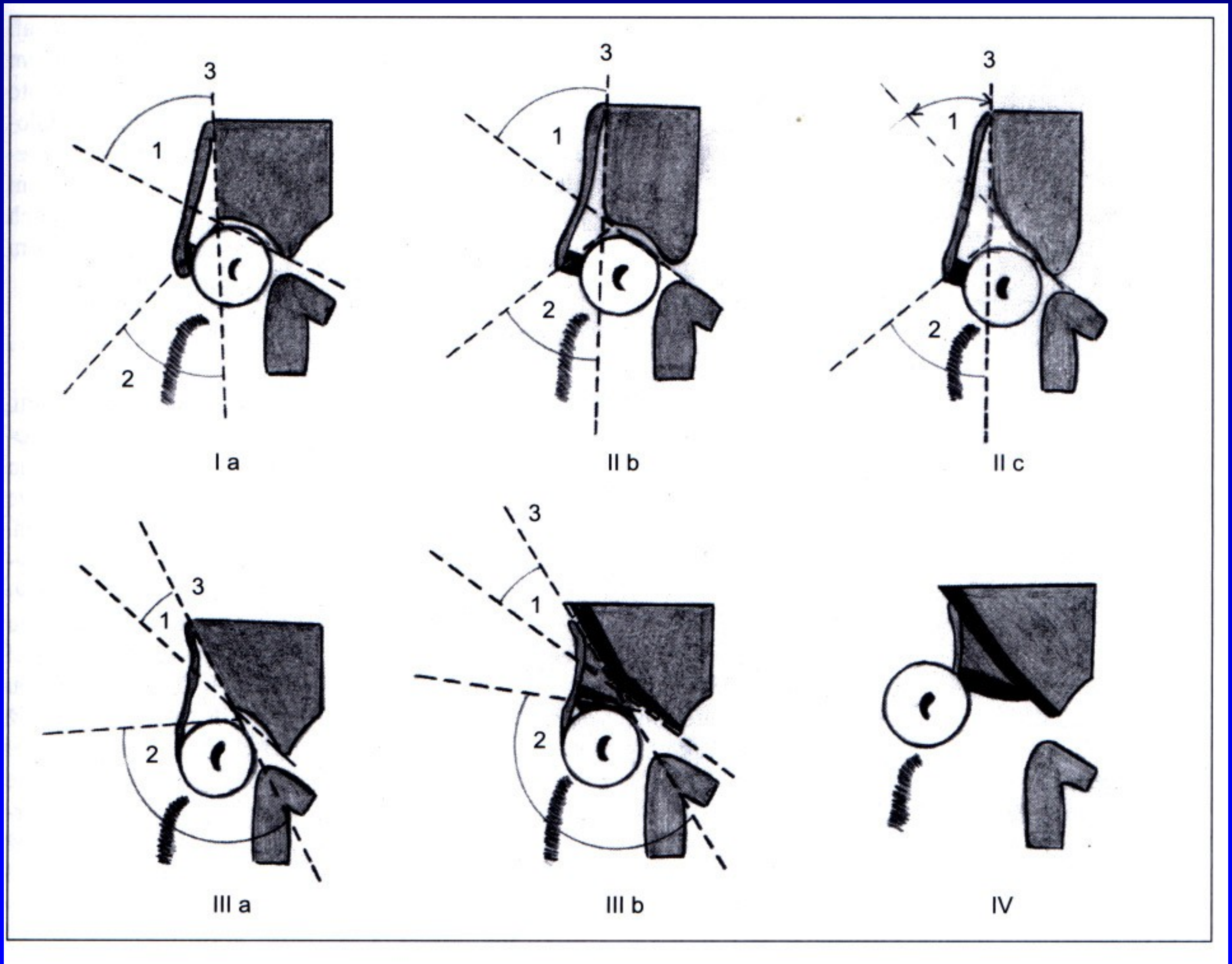
Obr. 71

1- iliac bone
2- cartilago of lateral acetabulum
3- labrum acetabulare
4- joint capsule
5- femoral head
6- osseous tectum
7- inferior margin of the iliac bone

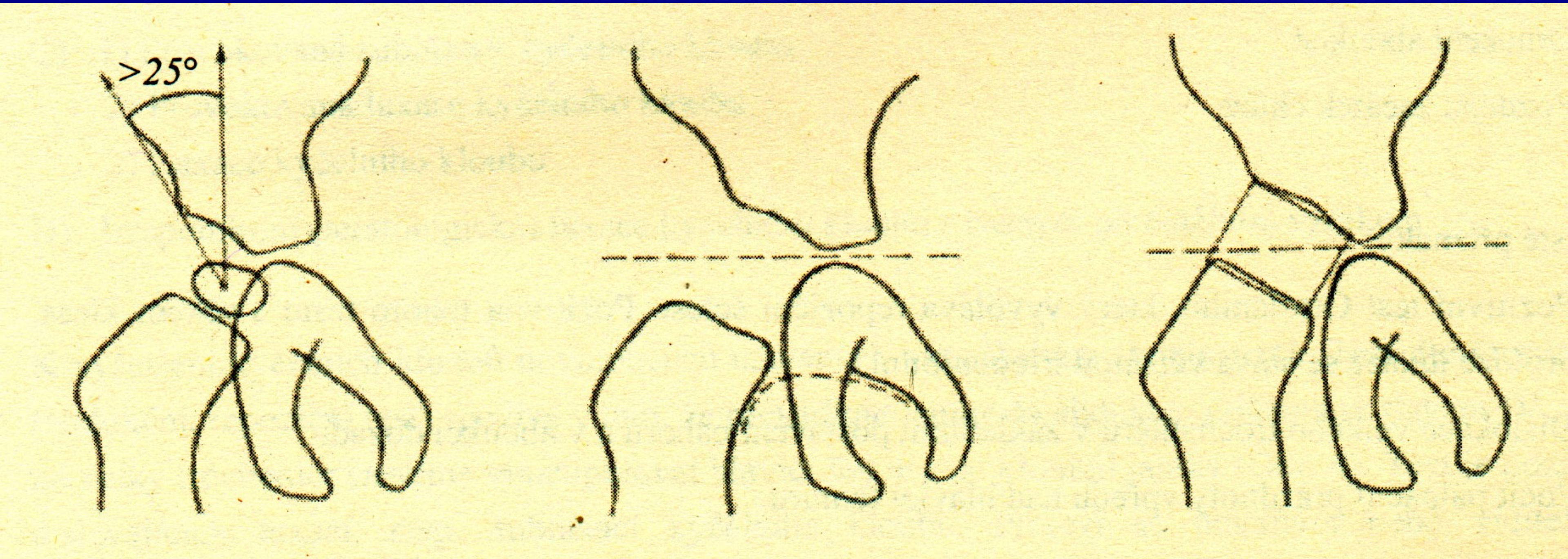
Graf classification- ultrasonography

Ia, Ib normal

IIa,b,c,d   dysplasia
IIIa,b   subluxation
IV   dislocation



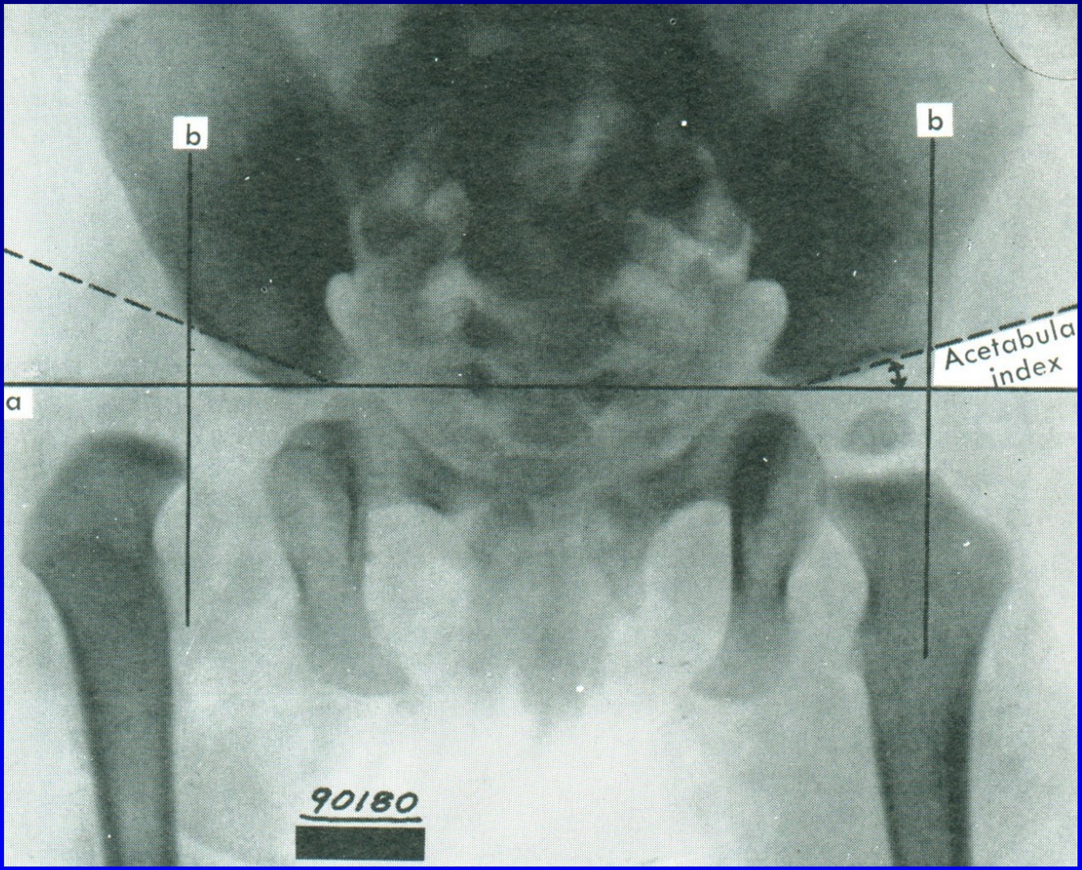
X-ray



Obr. 72

Wiberg angle Shenton line Kopizt paralerogram

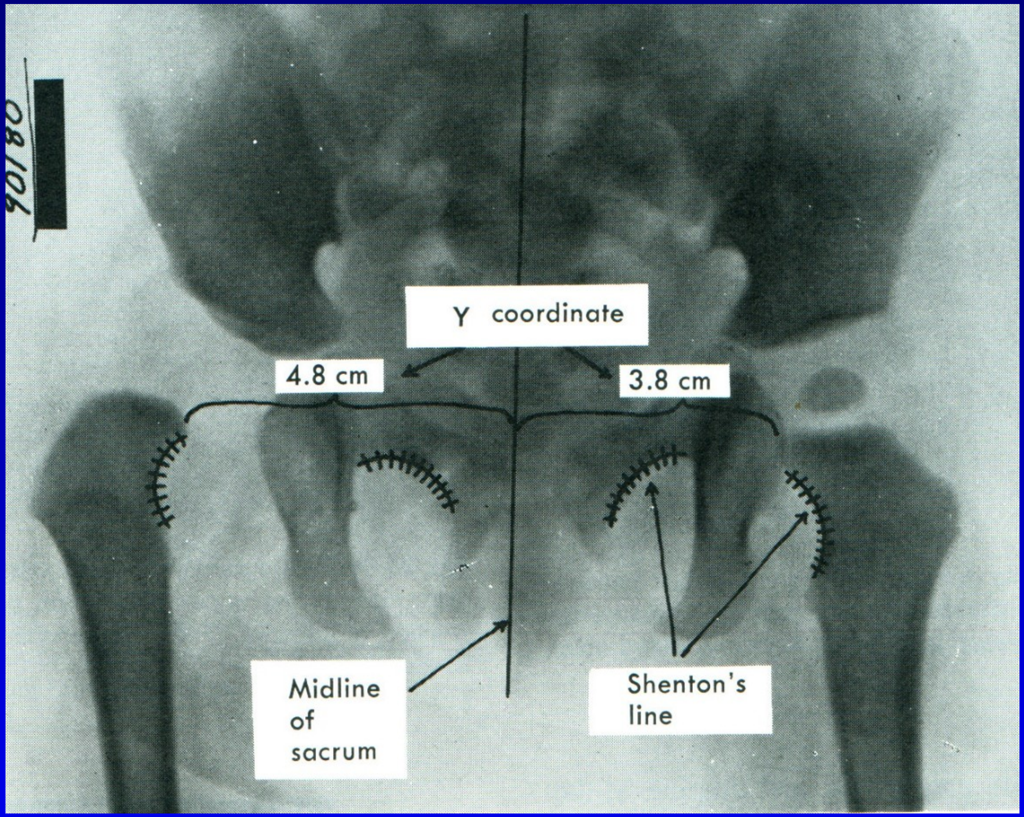
X-ray



Obr. 73

Dislocation of the right hip
Absent ossification of proximal epiphysis

X-ray



Obr. 74

Dislocation of the right hip
Femur is upwards and lateral
Shenton line is disrupted
No ossification of proximal epiphysis

Conservative management
- closed reduction

In the first month: reduction by gentle Ortolani manevuer- keep in abduction in Pavlik harness
Keep in safe zone: 90-120^{o} flexion
50-70^{o} abduction

Spontaneous reduction: up to 2- 3 months
- abduction positioning
- Frejka pillow
- Pavlik harness

Functional treatment

Obr. 75

Frejka pillow

Obr. 76

Pavlik harness

Over head traction

1. 2 weeks horizontal

2. 4 weeks in fexion of 100º
   gradually increasing of abduction
   up to 70º
   buttock is slightly above the bed

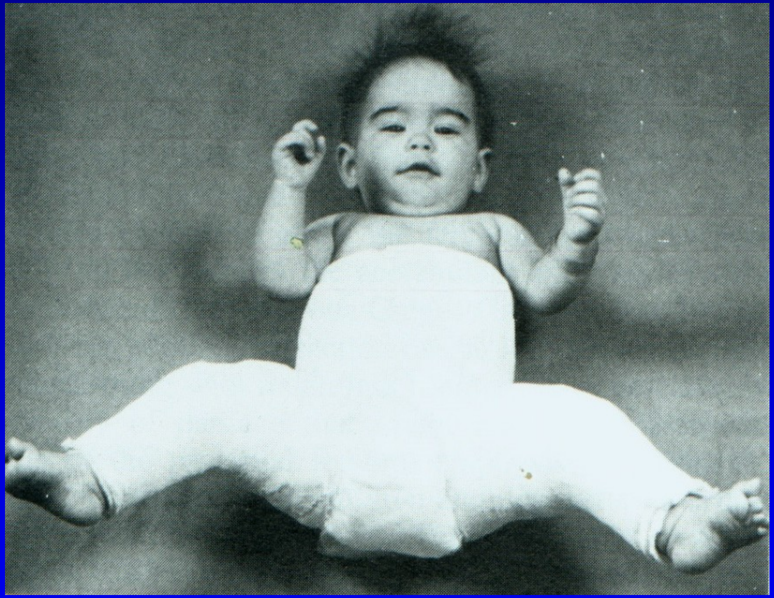
3. Bilateral hip spica
   flexion 100º, abduction 50º
   for 6 weeks

4. Aftertreatment with Pavlik harness
   of abduction apparatus

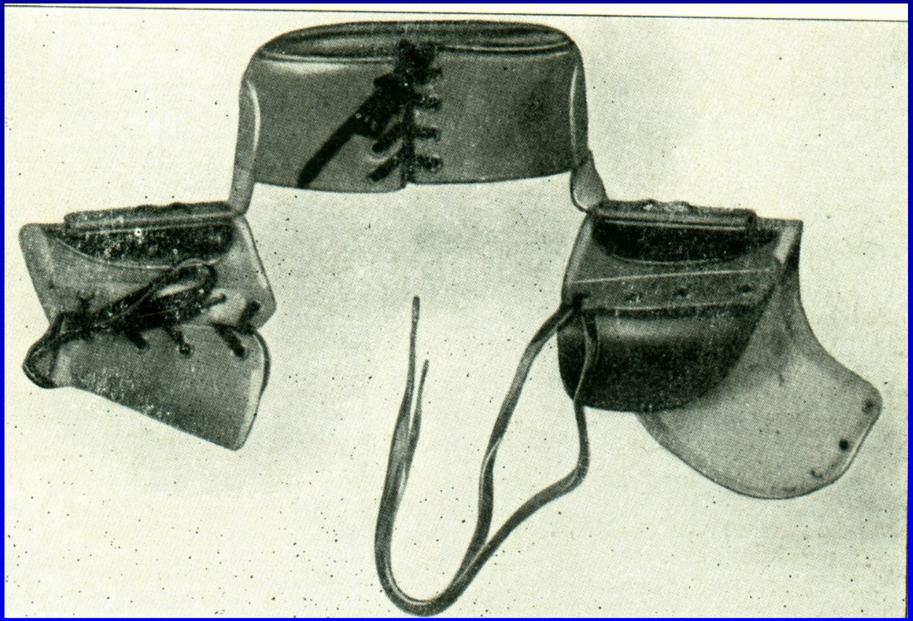
Obr. 77

Hip spica

Abduction apparatus



Obr. 78



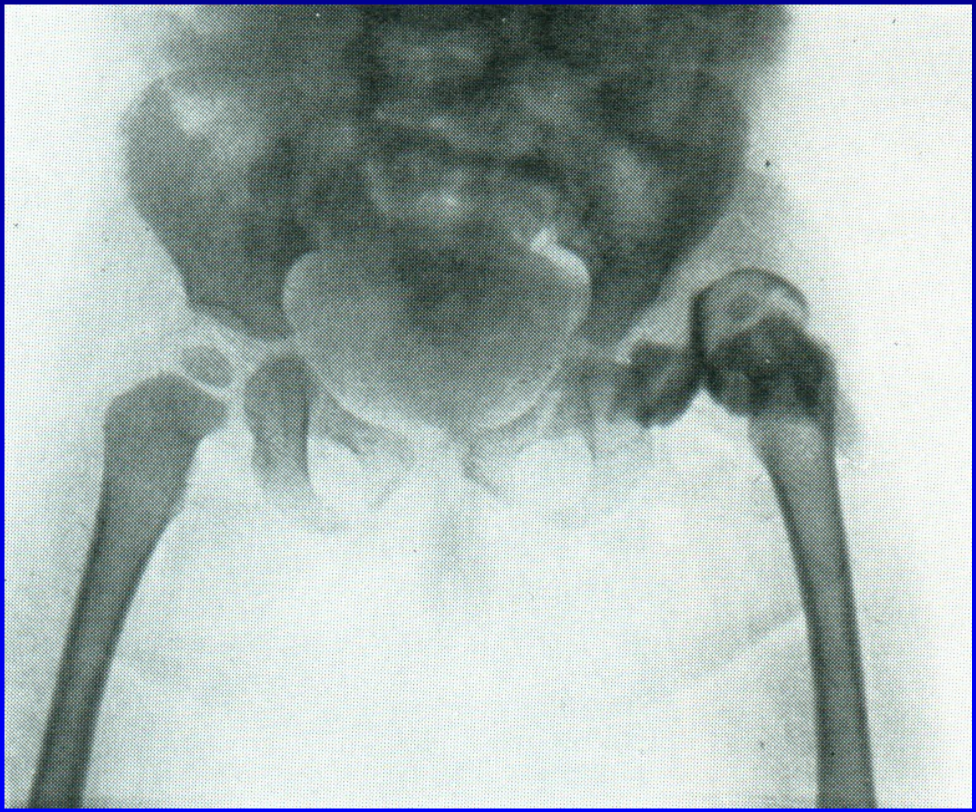
Obr. 79

Arthrography or MRI

Left hip

Inverted limbus

Constriction of joint capsule

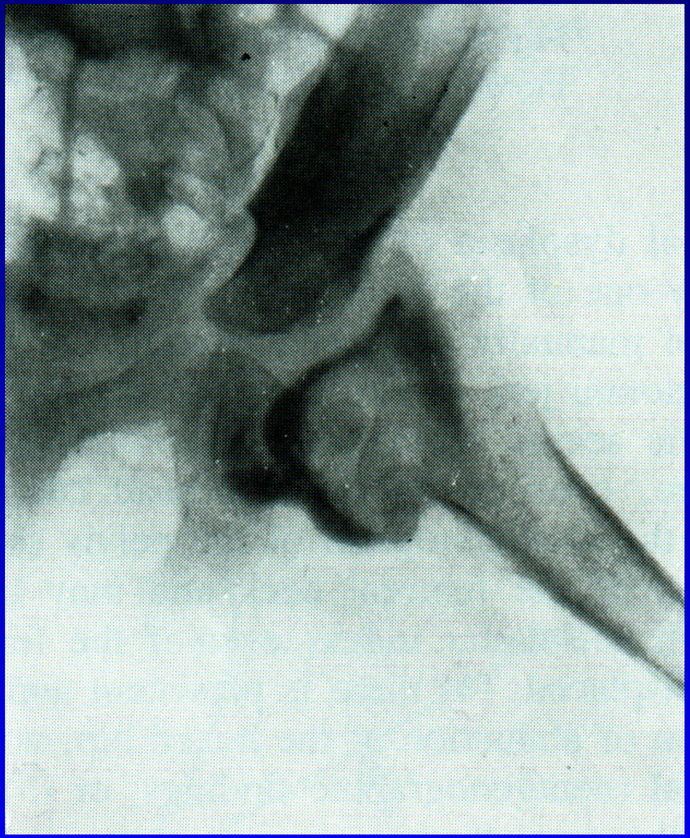


Obr. 80

Arthrography

Large inverted limbus

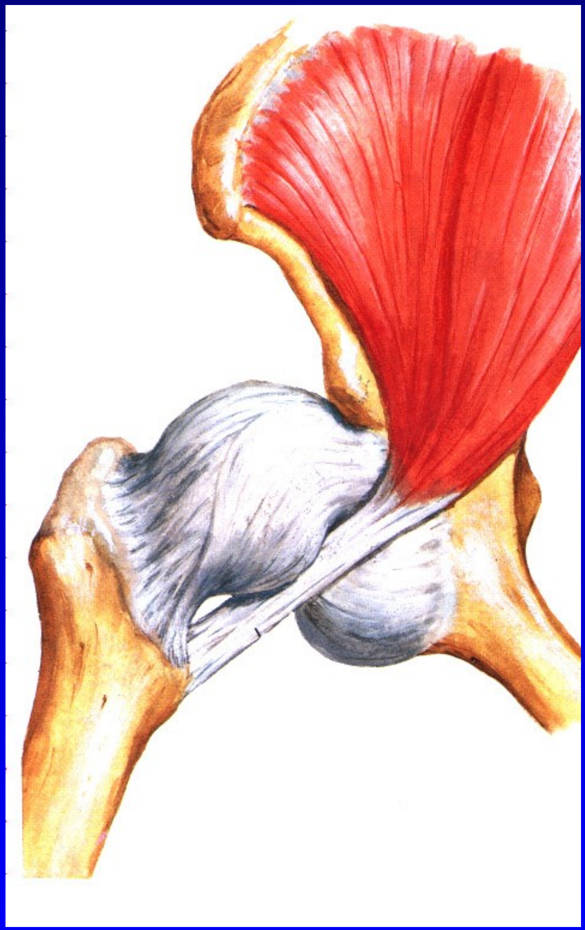
Dichotomy of femoral head



Obr. 81

Obstacles for reduction

1. Inverted limbus
2. Constriction of joint capsule
3. Hypertrophy of lig. capitis femoris
4. Ileopsoas tendon
5. Big anteversion of the femoral neck

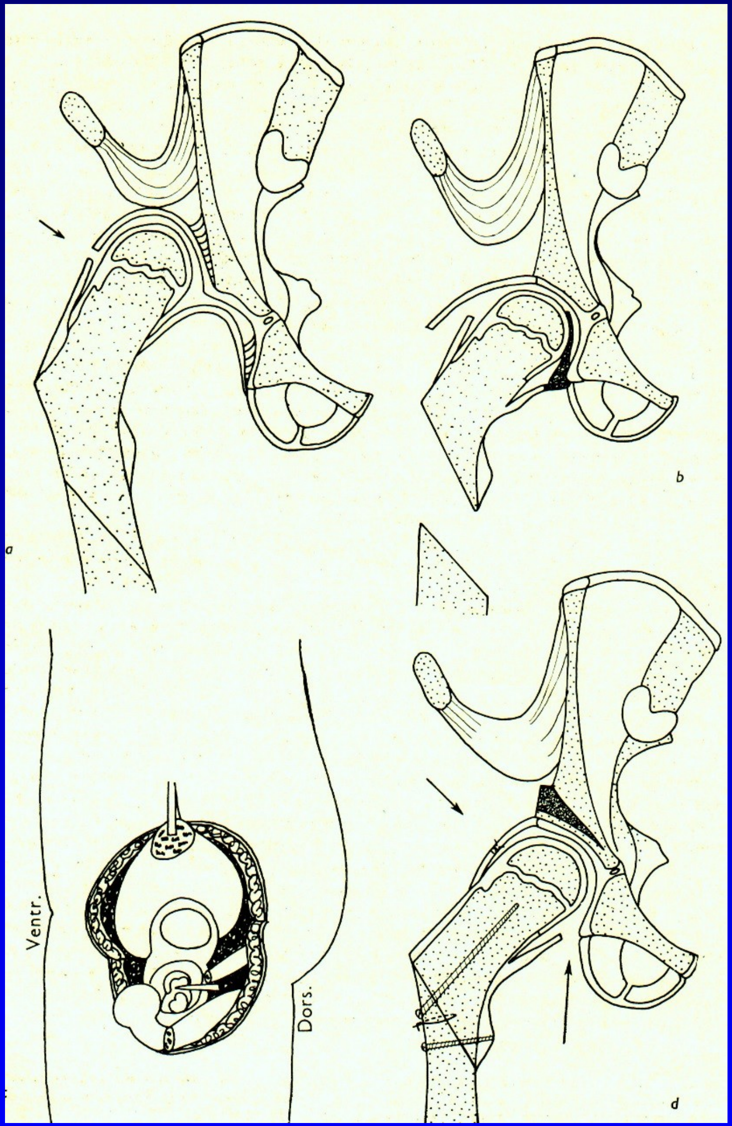


Obr. 82 Iliopsoas tendon

Operative treatment
Open reduction

Removal of the obstacle
Shortening osteotomy of the femur
Reduction into the acetabulum
Pelvic osteotomy-
Salter, Dega, Pemberton

Osteotomy of proximal femur
- CCD angle and anteversion



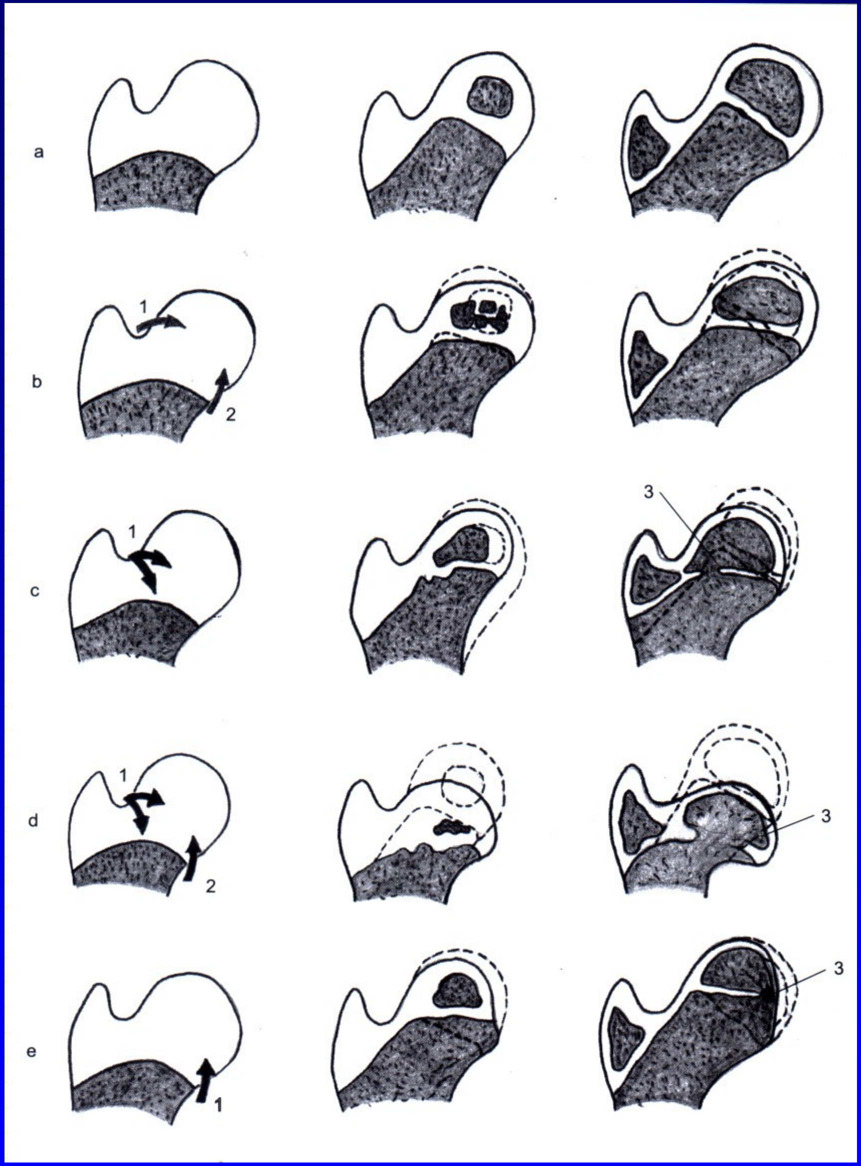
Obr. 83 Open surgery- Zahradníček

Salter osteotomy

Obr. 84

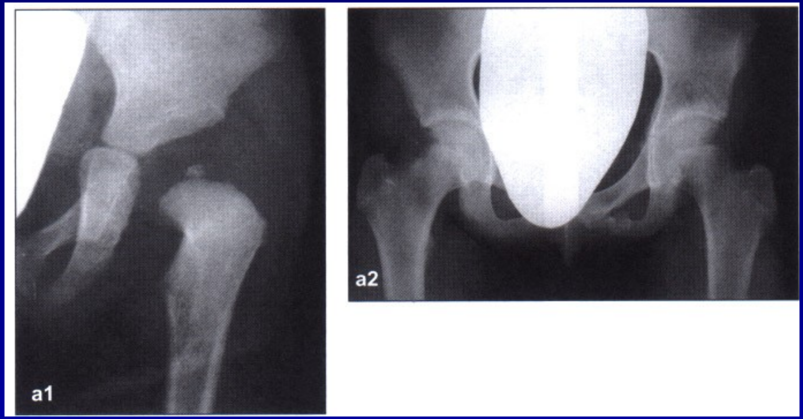
Ischemic necrosis in DDH

a   normal
b type I   slight flatening
c type II valgus deformity
d type III severe flattening,
             varus neck
e type IV medial part of
             epiphysis



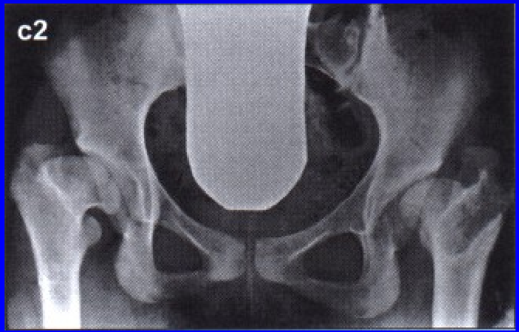
Buchholz and Ogden

Type I



Type II

Type III



Type IV.

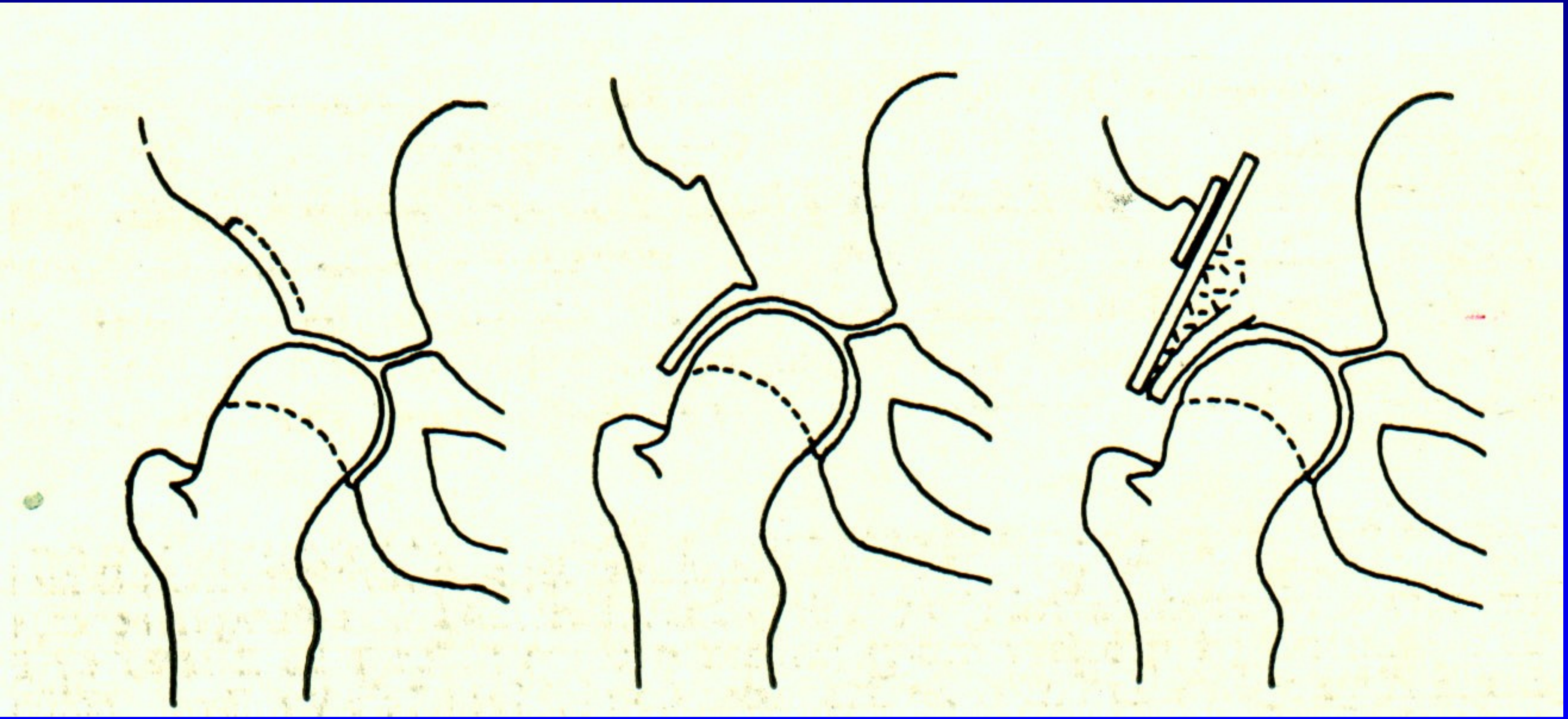
Operative treatment

Shelf plasty

Osteotomy of the pelvis
Steel, Sutherland, Eppright, Chiari

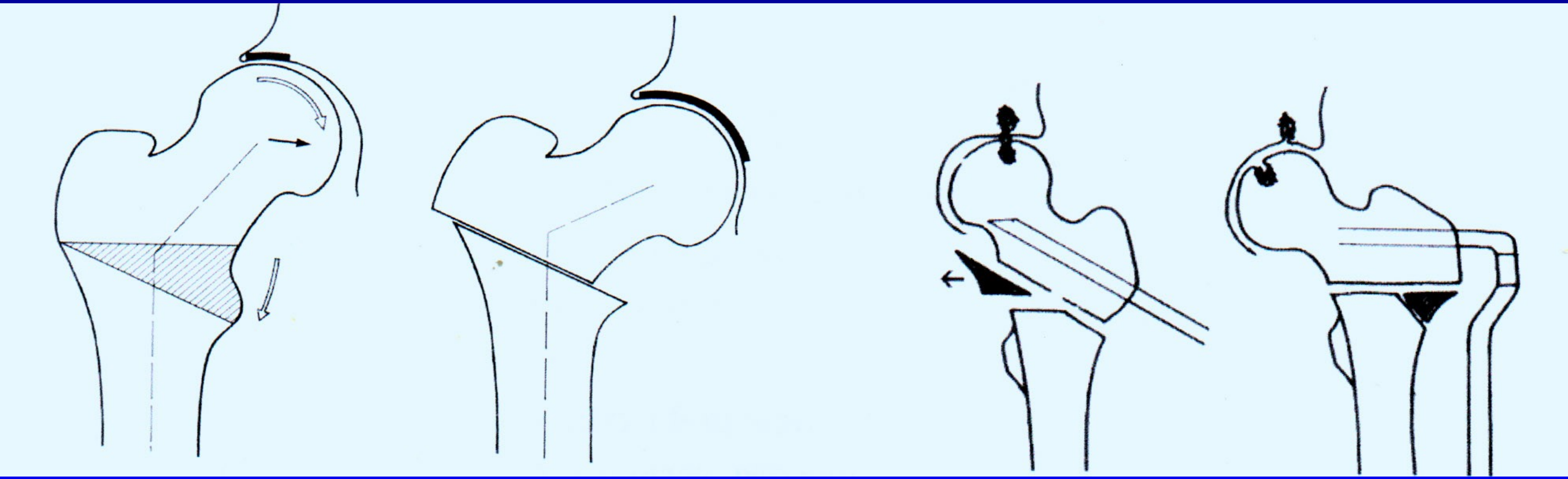
Osteotomy of the femur
- varus
- valgus
- shortening
- derotation

Shelf plasty



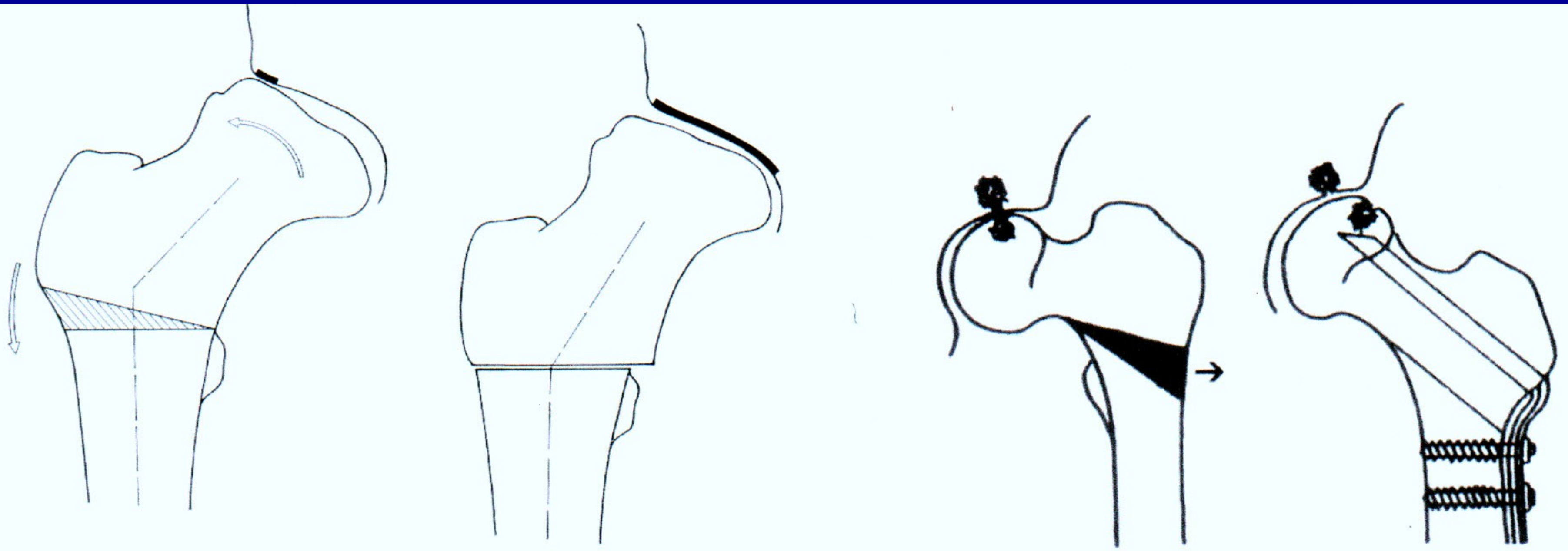
Obr. 87

Varus osteotomy



Obr. 85

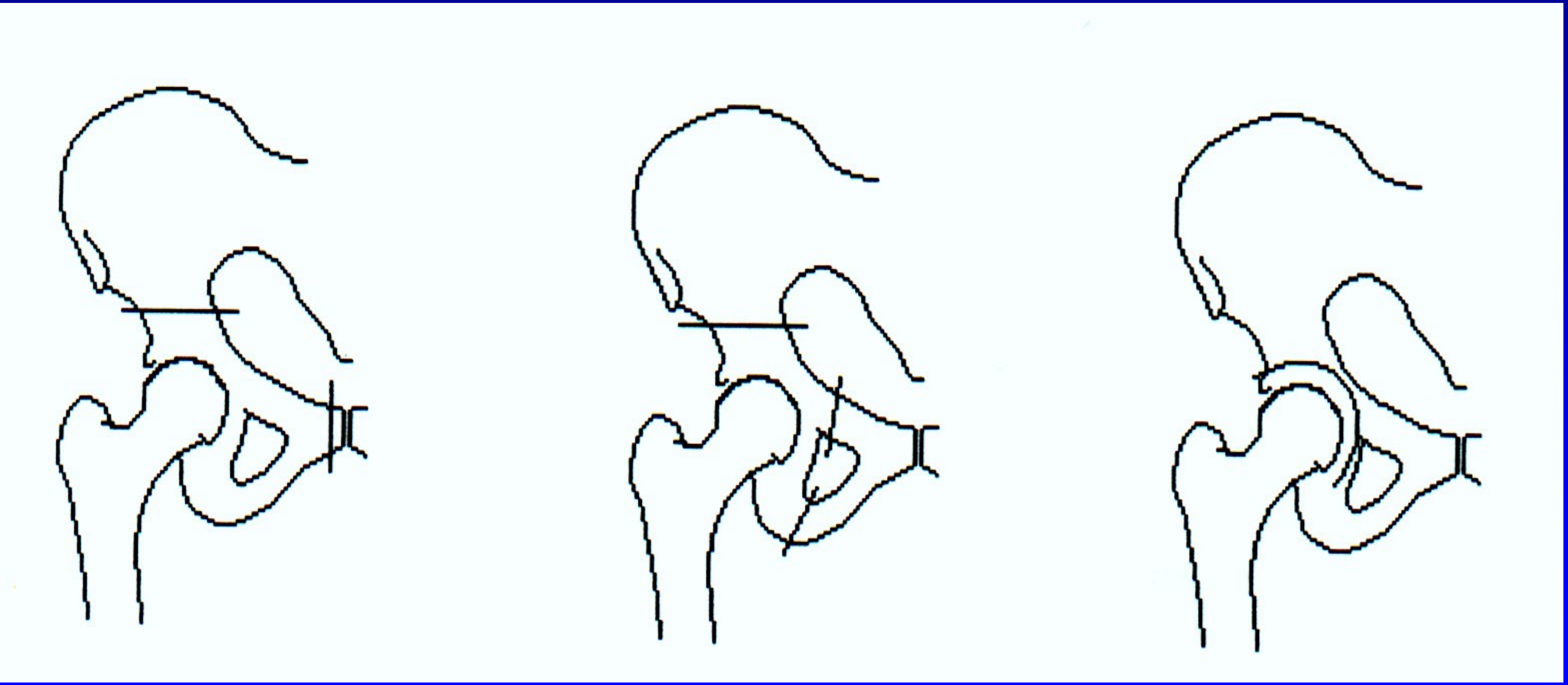
Valgus osteotomy



Obr. 86

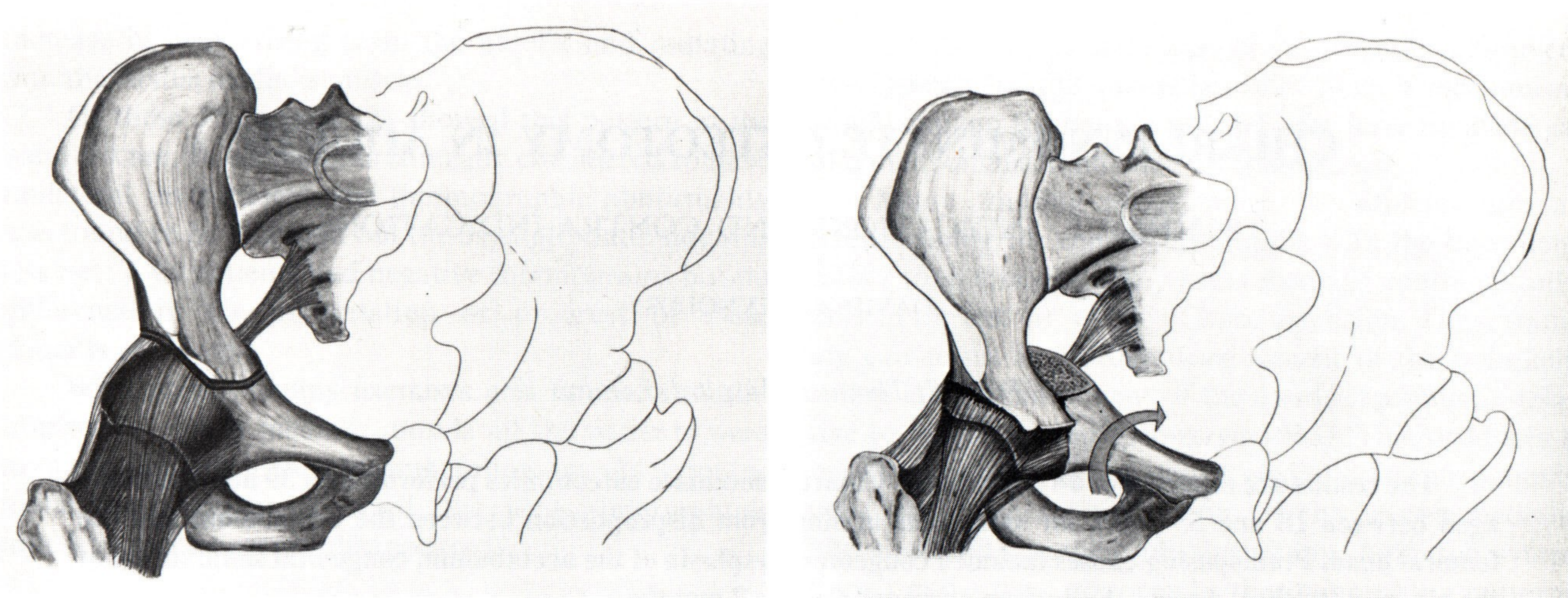
Osteotomy of the pelvis

Sutherland Steel Eppright



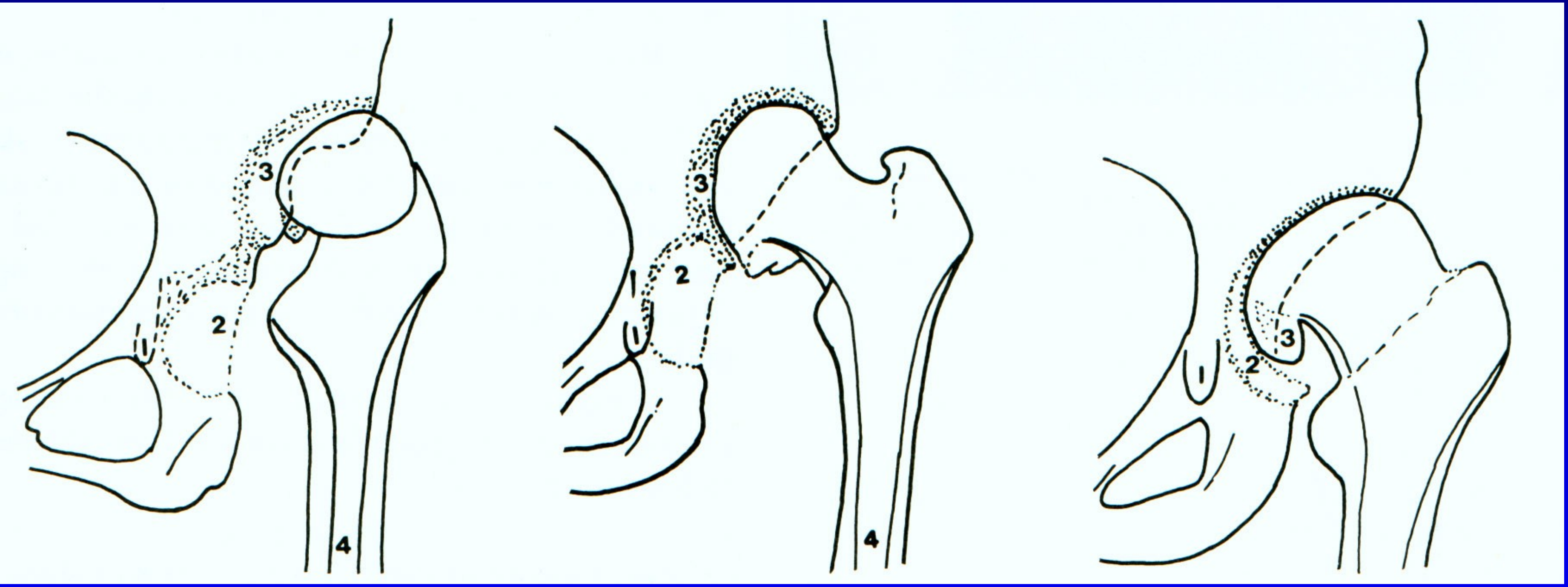
Obr. 88

Chiari osteotomy of the pelvis



Obr. 89

Consequences of DDH in adults



Obr. 65

Dislocation Subluxation Dysplasia

Consequences of DDH

Ischemic necrosis of femoral head

Residual deformity

Damage of the labrum

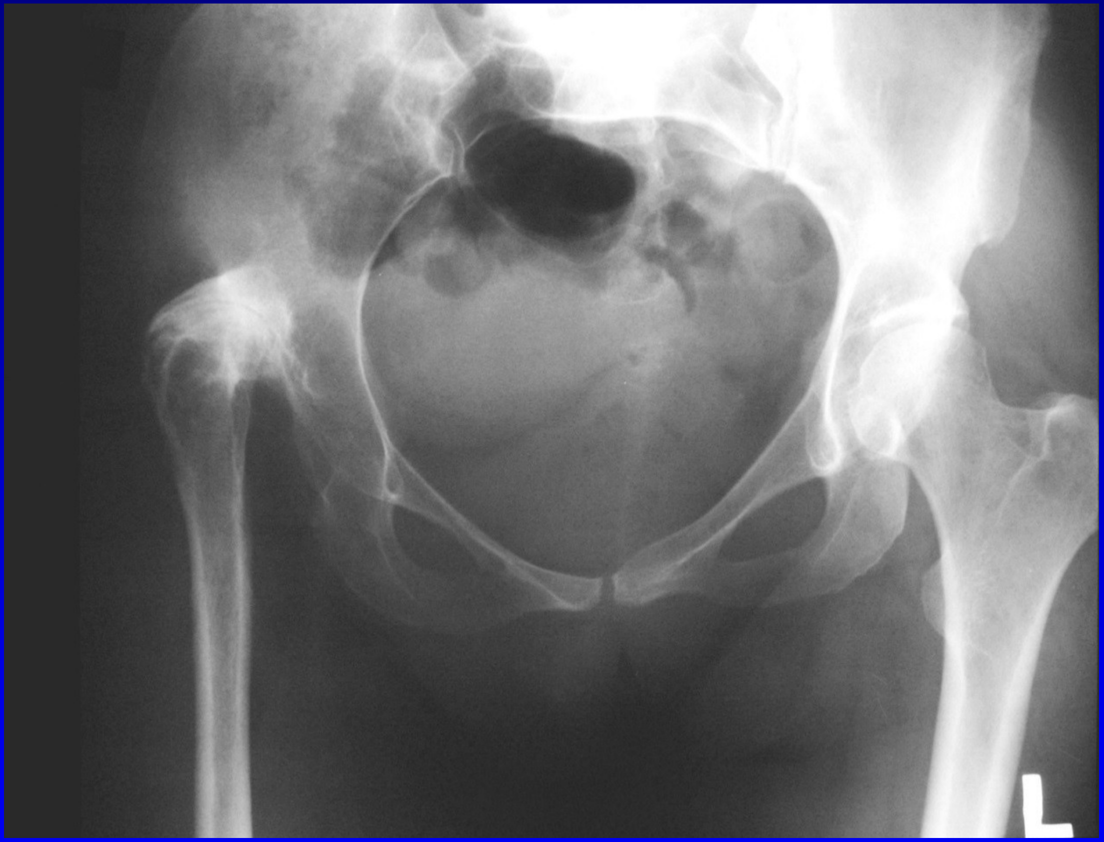
Obr. 90

Consequences of DDH

Shortening of the extremity
Pain
Limited movements
Limping
Weak muscles arround the hip
Dysplastic O.A.
Dyscomfort
Walking aids

Obr. 91

Consequences of DDH



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