

Cas de la semaine # 27

20 MARS 2017

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Histoire Clinique

- Sans particularité



62 ans

Radiographies des genoux



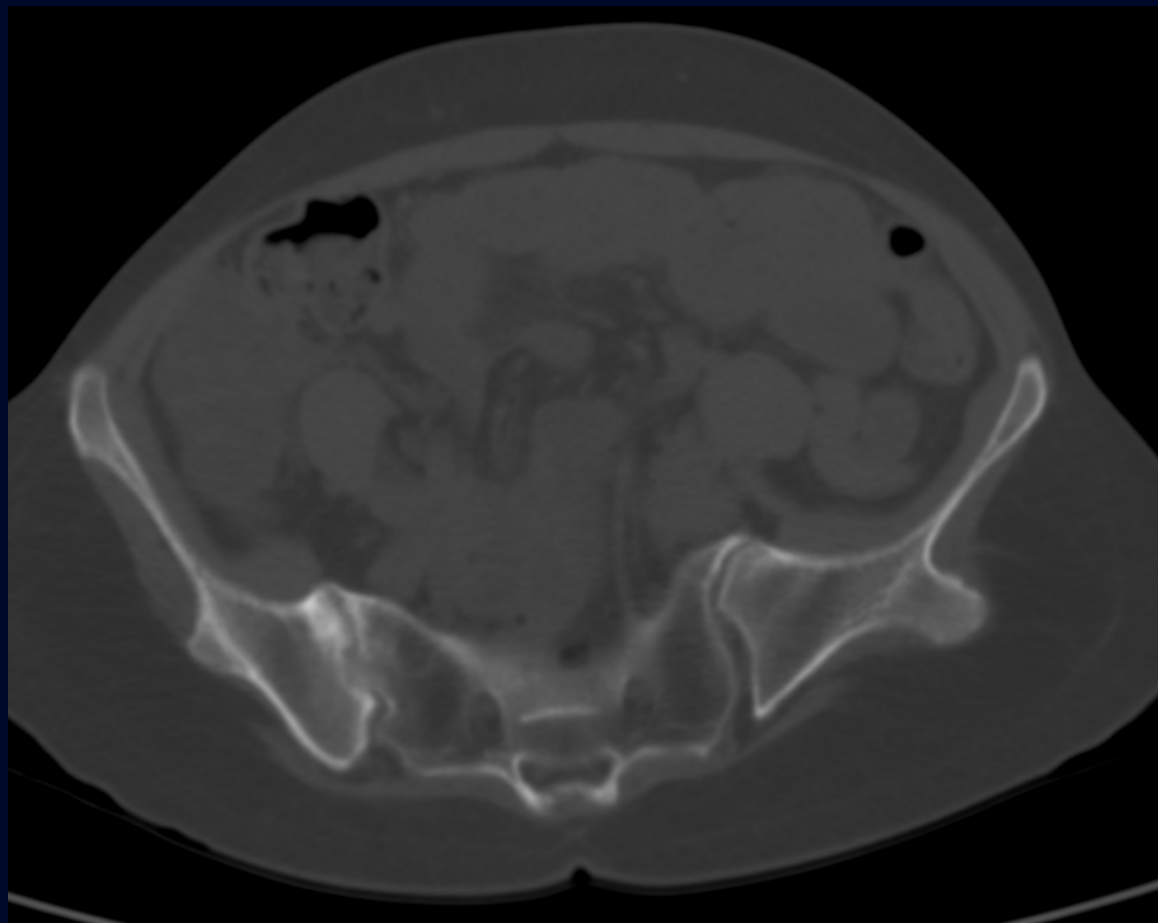
Radiographies des genoux



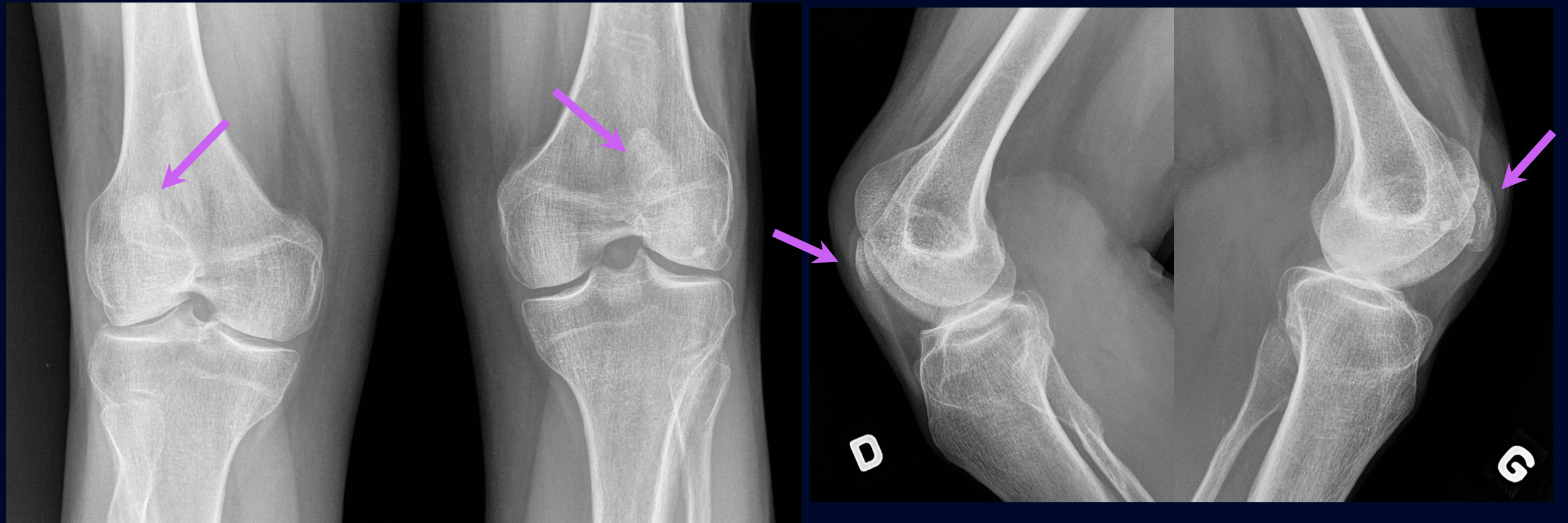
Bassin



CT scan du bassin



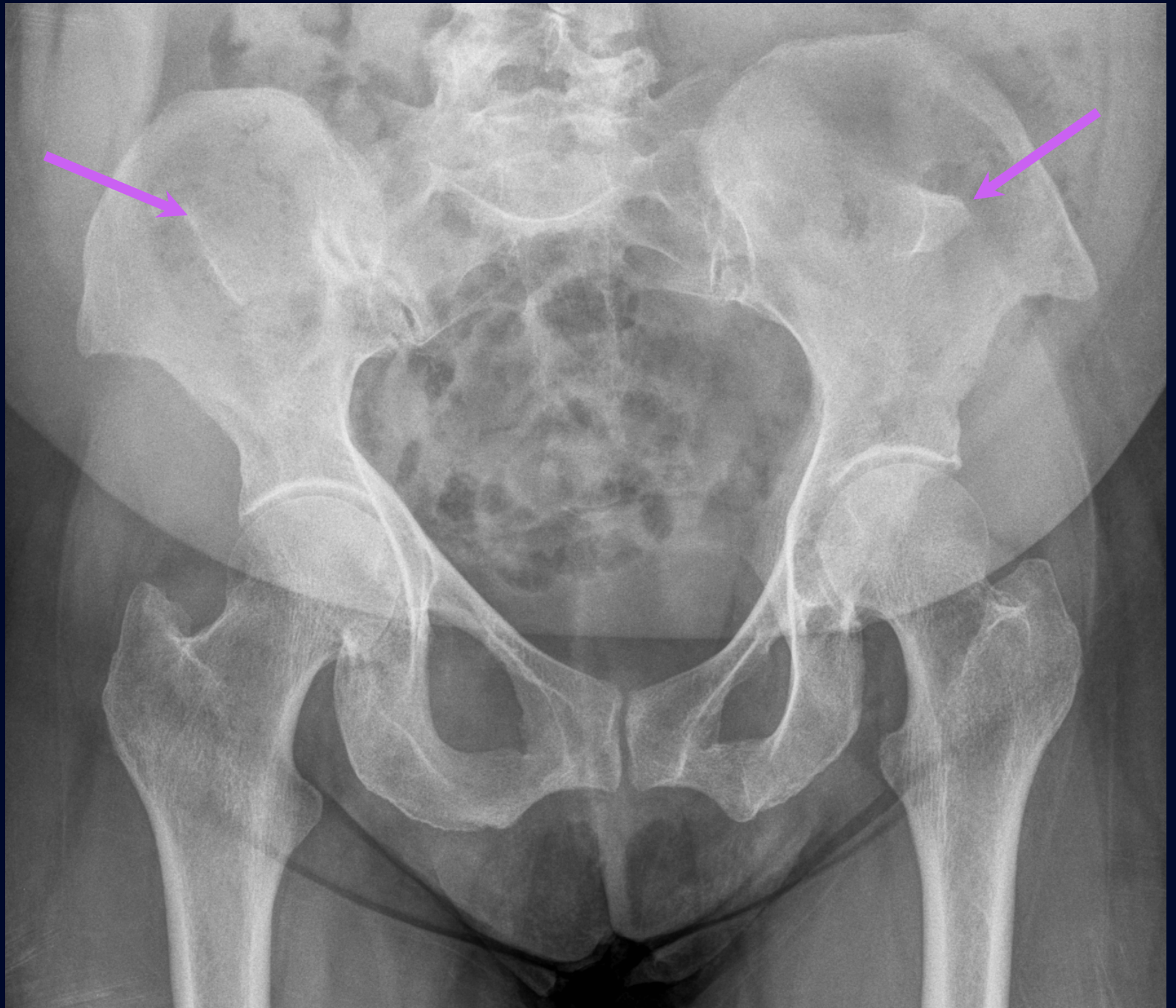
Radiographies des genoux



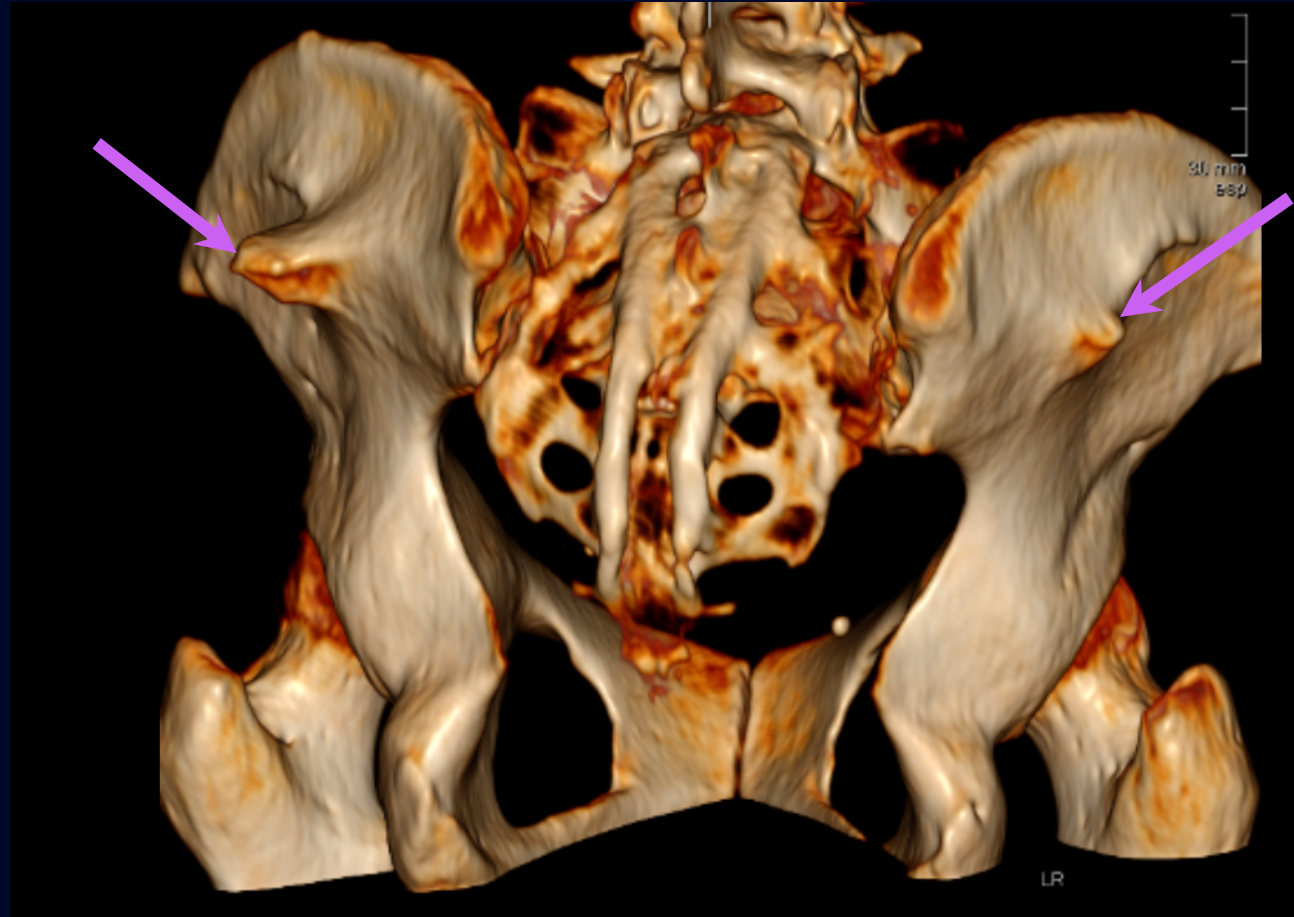
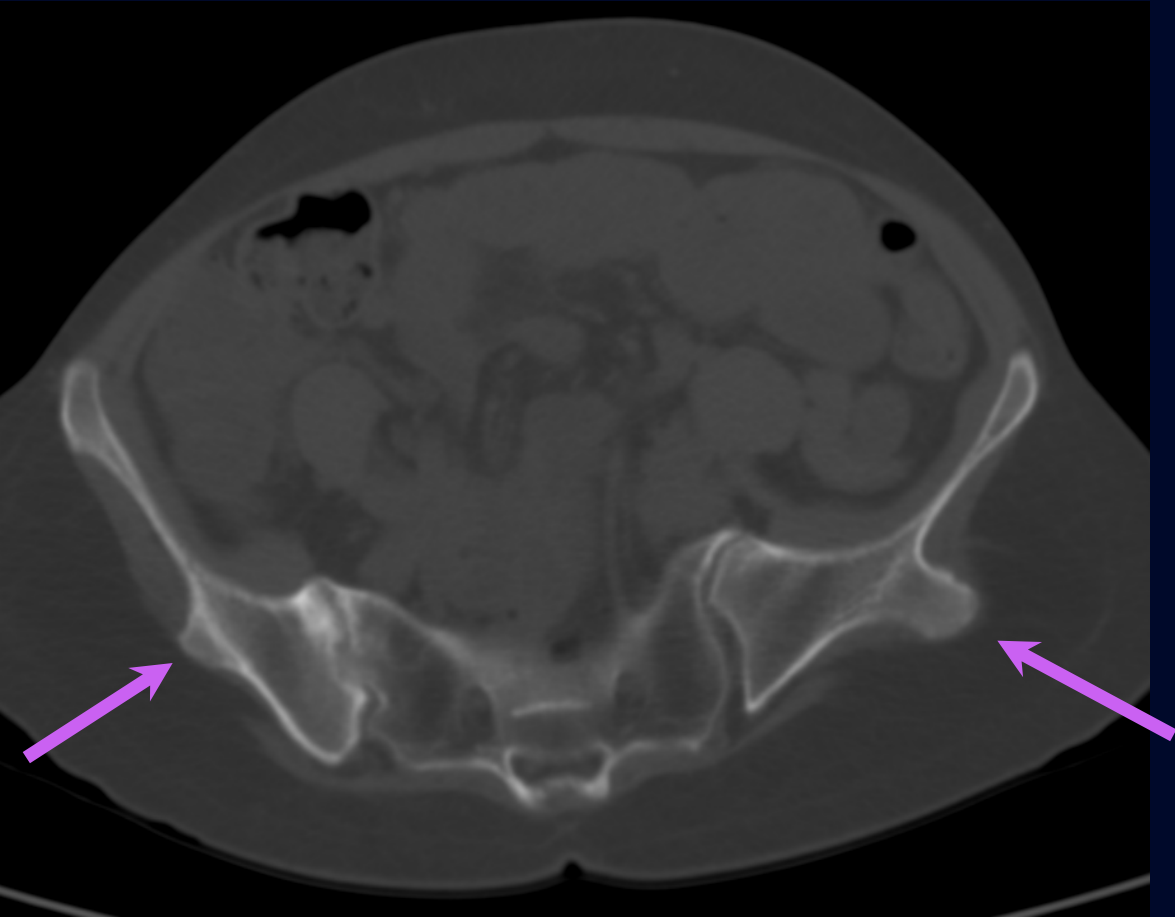
Hypoplasie patellaire bilatérale

Bassin

Présence de cornes
iliaques bilatérales



CT scan du bassin



Cornes iliaques postérieures bilatérales

Diagnostic différentiel

Absence / hypoplasie des rotules

- Chirurgie
- Nail-patella syndrome
- Small patella syndrome
- Patella aplasia-hypoplasia
- Syndrome de Meier-Gorlin
- Genito-patellar syndrome
- Trisomie 8
- Coffin-Siris syndrome
- RAPADILINO syndrome

Diagnostic final

Nail-patella syndrome

Synonymes:

- maladie de Fong
- syndrome de Turner-Keiser
- onycho-ostéodysplasie
héréditaire

Nail-patella syndrome

- Fréquence: 1/50.000
- Transmission autosomale dominante
- Mutation du facteur de transcription LMX1B localisé sur le chromosome 9

Description typique :

- ✓ dysplasie des ongles
- ✓ hypoplasie patellaire
- ✓ altération coudes
- ✓ cornes iliaques
 - les cornes iliaques postérieures sont PATHOGNOMONIQUES

Nail-patella syndrome

MANIFESTATIONS OSSEUSES

- Patellas hypoplasiques ou absentes
- Cornes iliaques postérieures
- Épines iliaques antérieures protubérantes
- Hypoplasie de la tête radiale ou du capitellum +/- luxation



**Tiré de
Réf. 5**

Dysplasie de la tête radiale (flèche), capitellum (*) et épicondyle latéral (tête de flèche)

AUTRES MANIFESTATIONS

- **Ongles:** absents, hypoplasiques ou dystrophiques



Tiré de réf. 1

- **Reins:** protéinurie chez 30-50%, progressant en insuffisance rénale chez 5-30%
- **Yeux:** fréquence accrue de glaucome à angle ouvert

Diagnostic différentiel du Nail-Patella Syndrome (adapté de Réf 1)

Syndrome	Similarities	Differences
Small patella syndrome (ischiopatellar dysplasia, coxo-podo-patellar syndrome)	Small or absent patellae Recurrent patella dislocations Pelvic anomalies	Defective ossification at the ischiopubic junction Ischial hypoplasia Infra-acetabular "axe-cut" notch No nail changes, No elbow changes No renal involvement, No ocular involvement
Patella aplasia-hypoplasia (PTLAH)	Isolated aplasia OR Hypoplasia of the patella	No nail changes , No elbow changes No renal involvement , No ocular involvement
Meier-Gorlin syndrome	Absent patellae Dislocation of the radial head	Microtia , Markedly short stature Delayed bone age Characteristic facial appearance Autosomal recessive inheritance
Genitopatellar syndrome	Absent patellae Renal anomalies Flexion deformities of the knees and hips Club foot	Hypoplasia of the ischia and iliac bones Genital anomalies Facial dysmorphism , Microcephaly , Intellectual disability Structural (multicystic kidneys or hydronephrosis) rather than functional abnormalities , Renal manifestations
Trisomy 8 mosaicism	Absent or hypoplastic patellae Limited elbow supination Abnormal nails	Significant learning difficulties Variable facial dysmorphism Camptodactyly and progressive joint restriction, usually of the fingers and toes
Coffin-Siris syndrome	Absence or hypoplasia of the nails and patellae Elbow dislocation	Nail hypoplasia, usually affecting the little finger nails Facial dysmorphism
RAPADILINO syndrome	Radial defects Absent or hypoplastic patellae Dislocated joints	Cleft palate , Facial dysmorphism , Short stature Radial defects, including absent or hypoplastic thumbs and radii Autosomal recessive inheritance

Nail-patella syndrome

Pronostic : Progresion vers l'insuffisance rénale terminale dans 5-30%

Traitement : symptomatique

- Analgésie et correction de l'atteinte osseuse selon symptômes
- Atteinte rénale
 - Surveillance annuelle pour hypertension et insuffisance rénale
 - Transplantation rénale au besoin
- Recherche de glaucome

Références

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