Mise au point sur les diagnostics étiologiques des acro ostéolyses

Acro-ostéolyses

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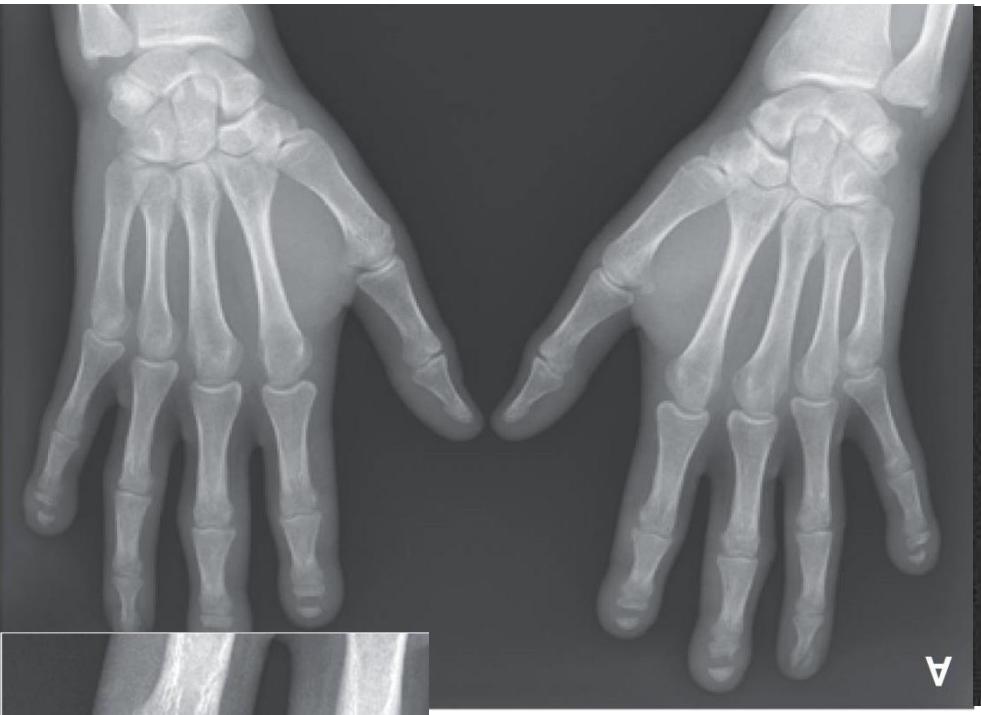
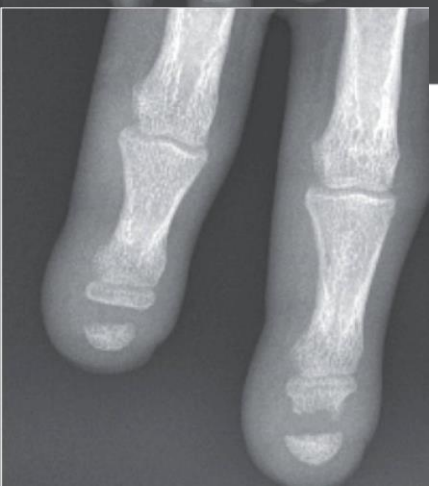
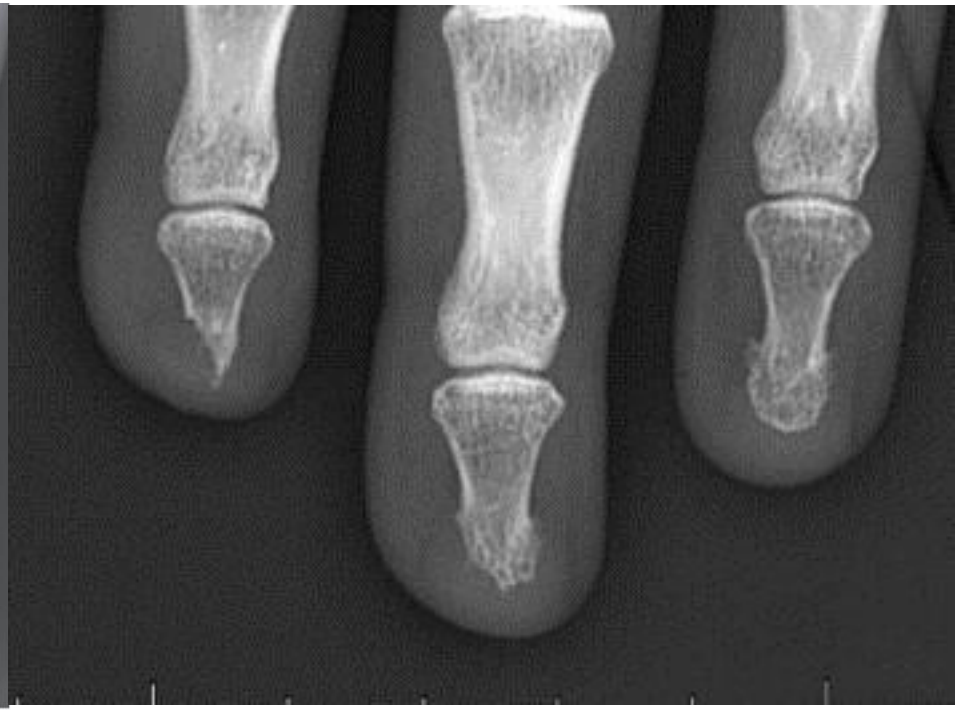
L'acro-ostéolyse de l'adulte est une destruction des phalanges distales des doigts ou des orteils, le plus souvent bilatérale.

AO transversale

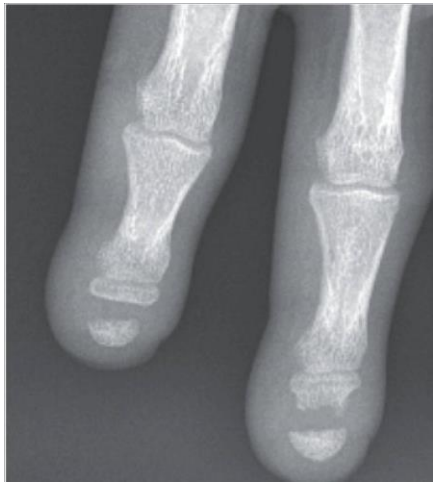




AO longitudinale



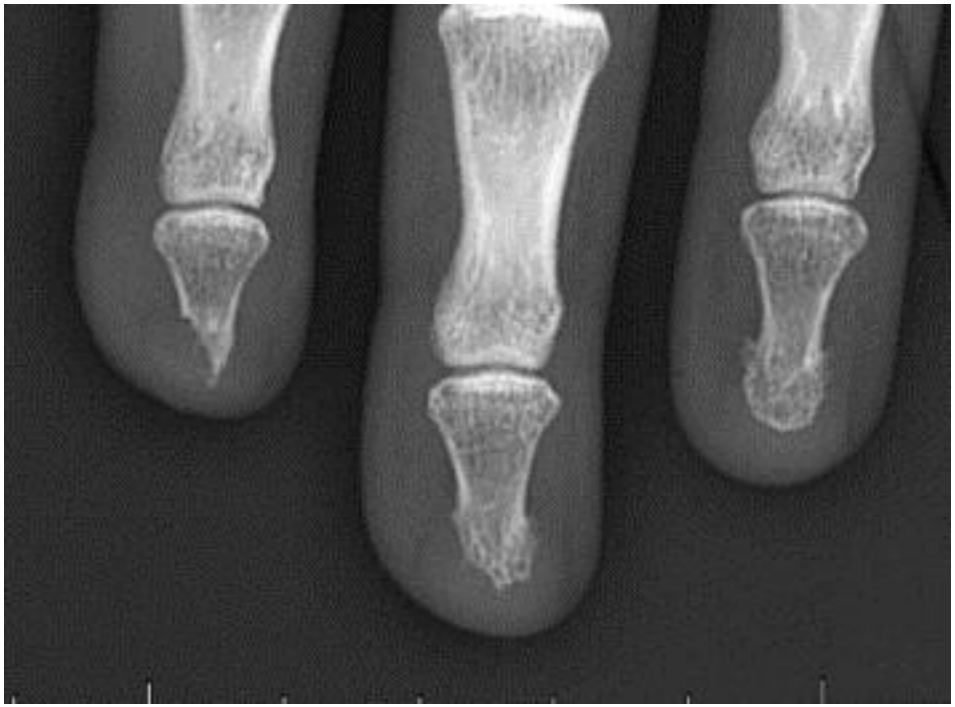
la base et la houppe
sont préservées



aspect de pseudo-fracture

Hyperparathyroïdie,
Intoxication au PVC
Syndrome d'Hadju et Cheney.

résorption concentrique de la houppe



Affections neurologiques
Affections vasculaires
Rhumatismes inflammatoires

GAMME DIAGNOSTIQUE

Acro-ostéolyse longitudinale

- Arthropathies nerveuses
- Sarcoïdose
- Goutte
- Tumeur
- Infection
- Médicaments
- Syndrome d'Ehlers-Danlos
- Réticulo-histiocytose multicentrique

Acro-ostéolyse transversale et/ou longitudinale

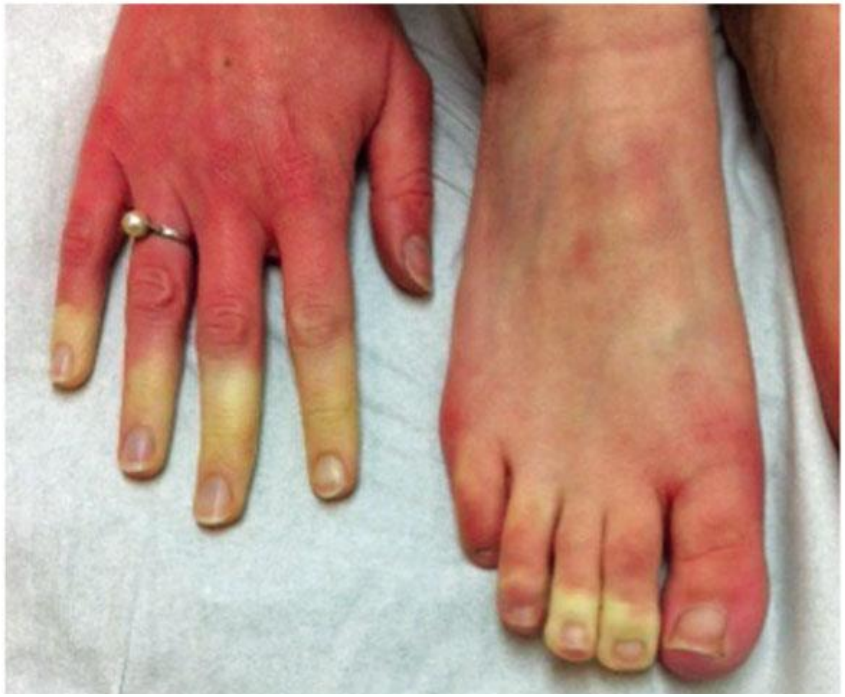
- Hyperparathyroïdie I ou II
- Rhumatisme psoriasique
- Polyarthrite rhumatoïde
- Traumatismes,
- Brûlures, gelures
- Sclérodermie, Polymyosite
- Venin de serpent ou scorpion
- Acro-ostéolyses idiopathiques

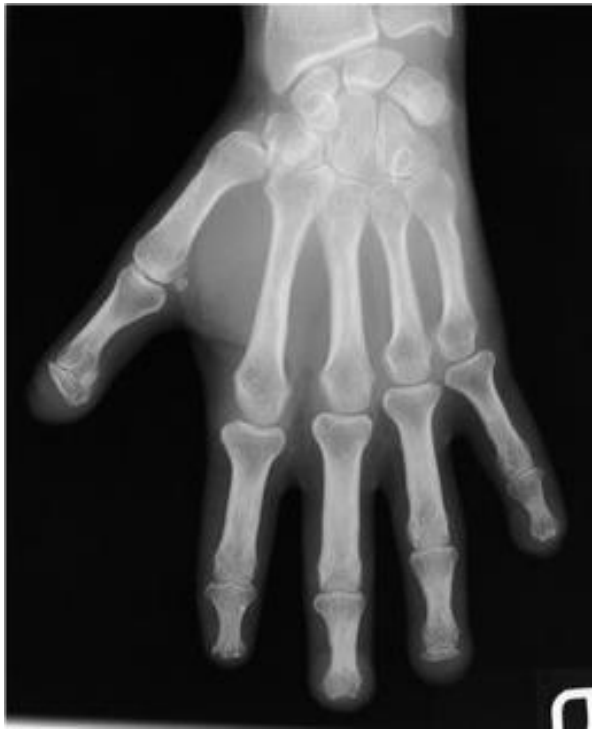
Acro-ostéolyse transversale

- Intoxication au chlorure de vinyle
- Pycnodysostose

Les acro-ostéolyses acquises

La sclérodermie et le syndrome de Raynaud





Prévalence de 20% selon les séries

ARTHRITIS & RHEUMATOLOGY
Vol. 68, No. 1, January 2016, pp 201–209
DOI 10.1002/art.39424

Association of Acroosteolysis With Enhanced Osteoclastogenesis and Higher Blood Levels of Vascular Endothelial Growth Factor in Systemic Sclerosis

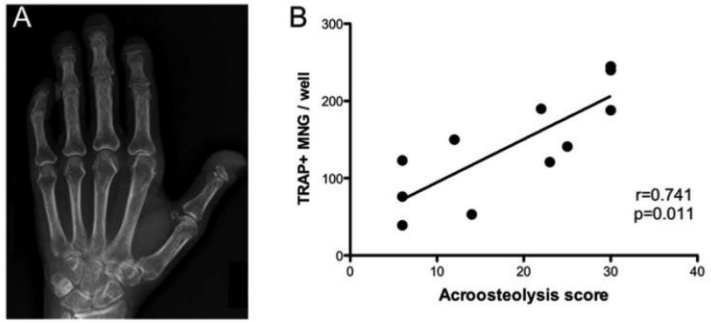
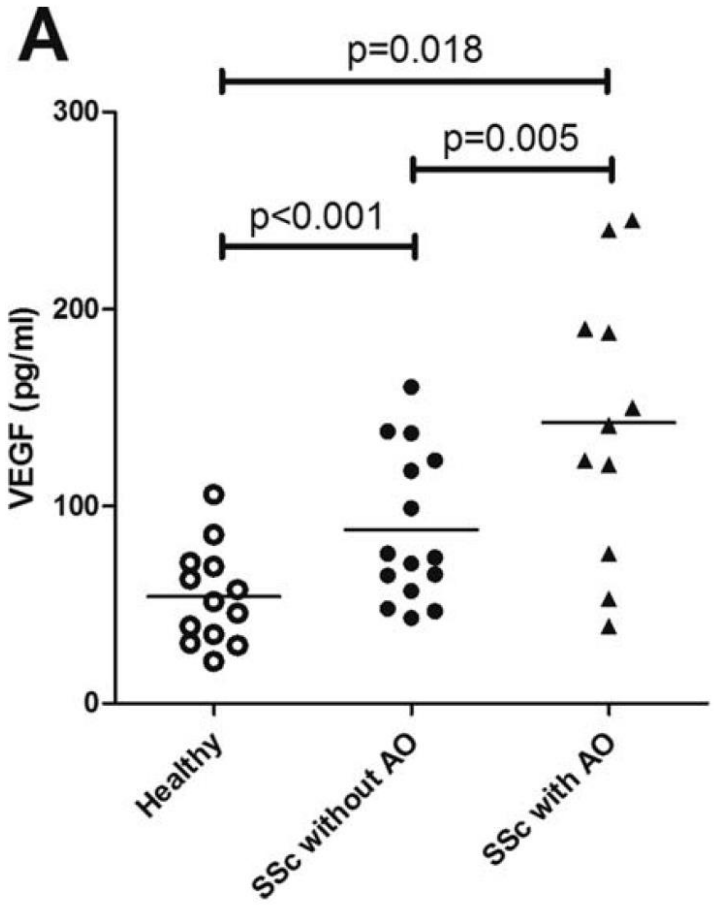


Figure 4. Association between the degree of acroosteolysis (AO) and osteoclastogenesis. **A**, Plain radiograph of the right hand of a patient with systemic sclerosis (SSc) with AO, revealing significant resorption of the distal phalanges. **B**, Linear relationship between the radiographic extent (severity) of AO and the number of tartrate-resistant acid phosphatase (TRAP)–positive multinucleated giant cells (MGCs [MNGs]) formed in each SSc patient. A composite AO score was generated by grading radiographic AO severity in each digit on a scale of 0–3, where 0 = none, 1 = minimal/doubtful, 2 = evident in less than 50% of the tuft, and 3 = evident in more than 50% of the tuft.



Hypoxie ⟶ VEGF ⟶ Ostéoclastogénèse

Les acro-ostéolyses acquises

Le syndrome de Raynaud

Acro-osteolysis

Ivo R Ferreira, Vital S Domingues

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A 71-year-old man hospitalised with tracheobronchitis, complained of hand discolouration. His hands showed the three-phases of skin colour changes (white, blue, and red) and a diagnosis of Raynaud's syndrome was established. When questioned about the first time he had these symptoms, the patient noted that they had been recurrent for about 20 years. He had relatively short fingers, particularly of the thumbs, and no bone was palpable in most of the distal phalanges (figure A). Radiography of his hands showed bone resorption of almost all terminal phalanges of both hands, so-called acro-osteolysis (figure B). Immunological investigations and clinical features excluded several diseases commonly associated with Raynaud's syndrome. Nailfold capillaroscopy was done and showed no changes, supporting the diagnosis of primary Raynaud's syndrome. The most common causes of acro-osteolysis include scleroderma, psoriatic arthritis, occupational causes, injury (eg, thermal burn), and hereditary syndromes (eg, Hadju-Cheney syndrome). In patients with long-standing primary Raynaud's syndrome, chronic vascular deficiency may lead to acro-osteolysis.

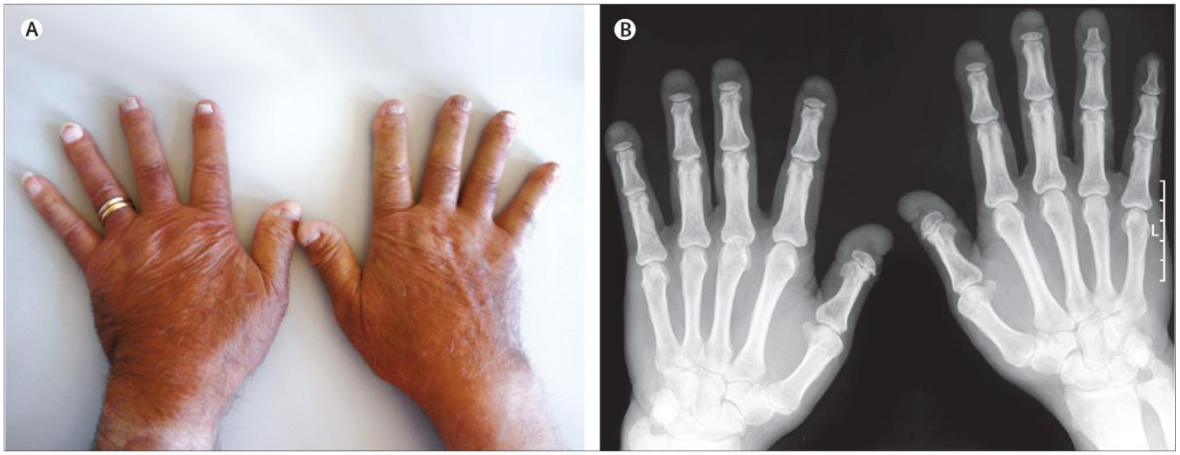


Figure: **Acro-osteolysis**
(A) Photo of the patient's hands; (B) radiography of the patient's hands showing acro-osteolysis of almost all terminal phalanges.

Les acro-ostéolyses acquises

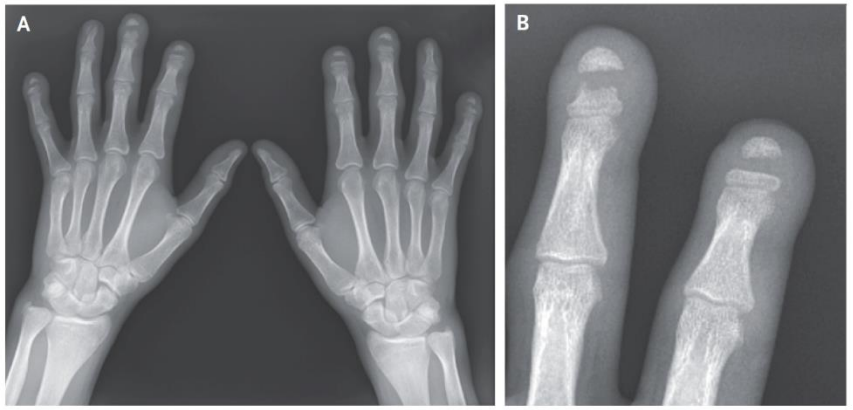
Le syndrome de Raynaud

The NEW ENGLAND JOURNAL of MEDICINE

IMAGES IN CLINICAL MEDICINE

Lindsey R. Baden, M.D., *Editor*

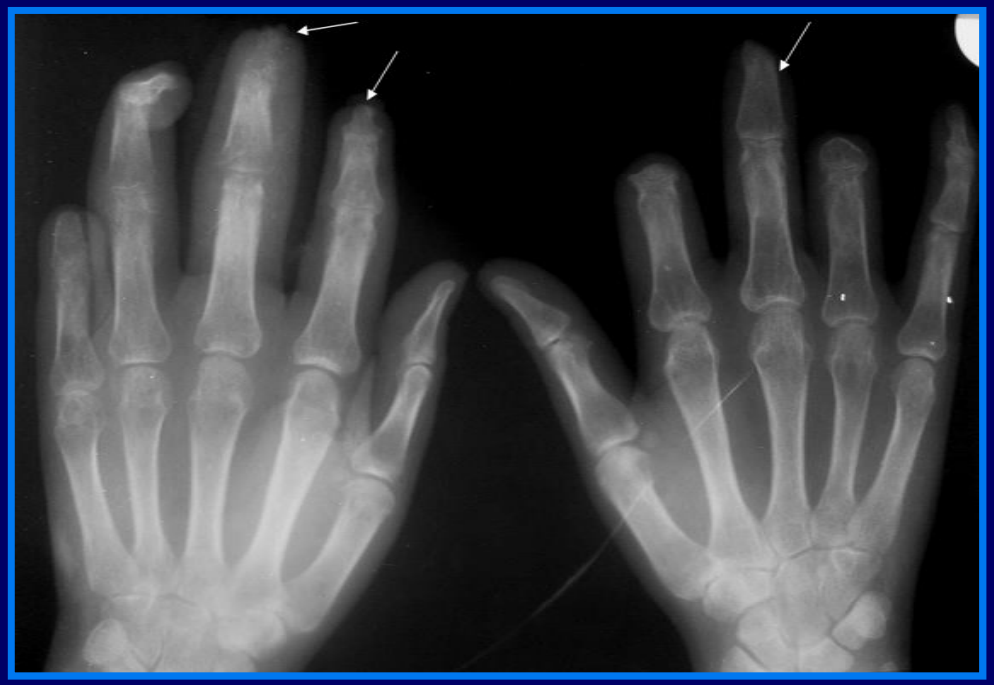
Band Acro-osteolysis



A 23-YEAR-OLD MALE CONSTRUCTION WORKER WITH A HISTORY OF AUTOimmune sclerosing cholangitis presented with a 3-year history of coldness affecting both hands, triphasic Raynaud's phenomenon, and tightening of the skin. Physical examination revealed mild clubbing of the fingers; patchy, nodular thickening of the skin on the hands and forearms; and linear plaques of fibrosis extending proximally past the elbows. His serum was positive for antinuclear antibody (titer, 1:1280) in a homogeneous pattern. Tests for anti–double-stranded DNA, anti-ribonucleoprotein, and anti-Scl-70 antibodies were negative. Plain radiographs of the hands showed sharply demarcated osteolysis of the midshaft of the distal phalanges of the index, middle, and little fingers on both hands and of the left ring finger (Panel A, with Panel B showing an enlarged image of the middle and index fingers of the left hand). This rare finding, known as band acro-osteolysis, has been associated with exposure to polyvinyl chloride (PVC) and with the Hadju–Cheney syndrome. The patient said he had had no exposure to PVC and had no personal or family history of congenital bone dysplasia. Because his symptoms were suggestive of atypical systemic sclerosis, immunosuppressive therapy with mycophenolate was initiated, and treatment with losartan was started for Raynaud's phenomenon. At 1 year, his condition was clinically and radiologically stable and the Raynaud's symptoms were reduced.

DOI: 10.1056/NEJMicm1311597

AFFECTIONS VASCULAIRES



THROMBO-ANGÉITE OBLITÉRANTE (MALADIE DE LEO BUERGER)

Jeune homme tabagique + Phénomène de Raynaud et ischémie périphérique du membre supérieur + acro-ostéolyses et amputations.

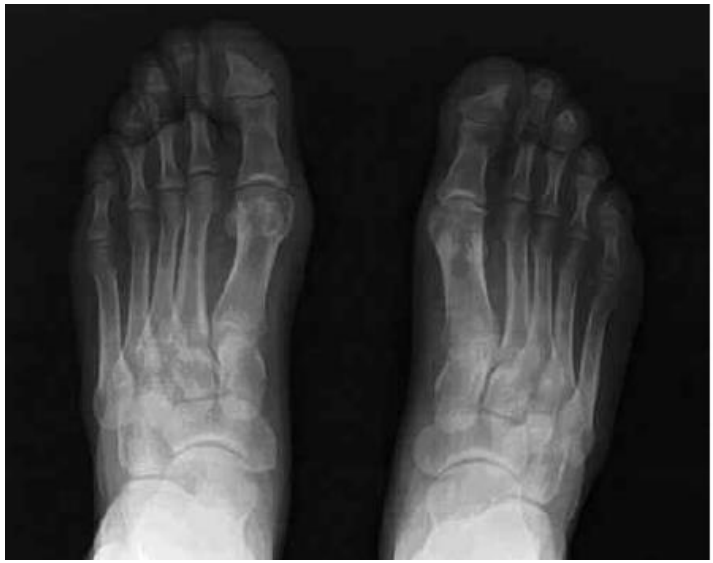
Les acro-ostéolyses acquises

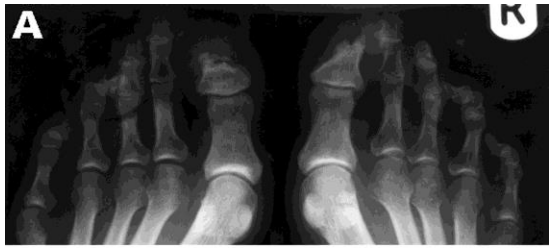
AO Toxique (PVC)

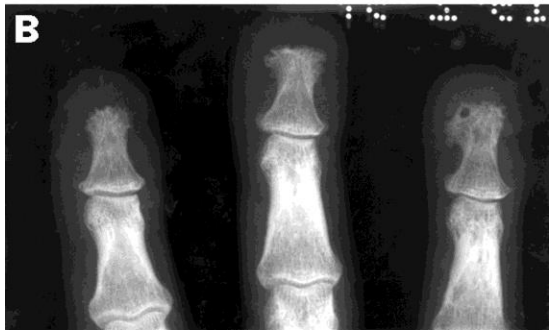
Rhumatisme psoriasique

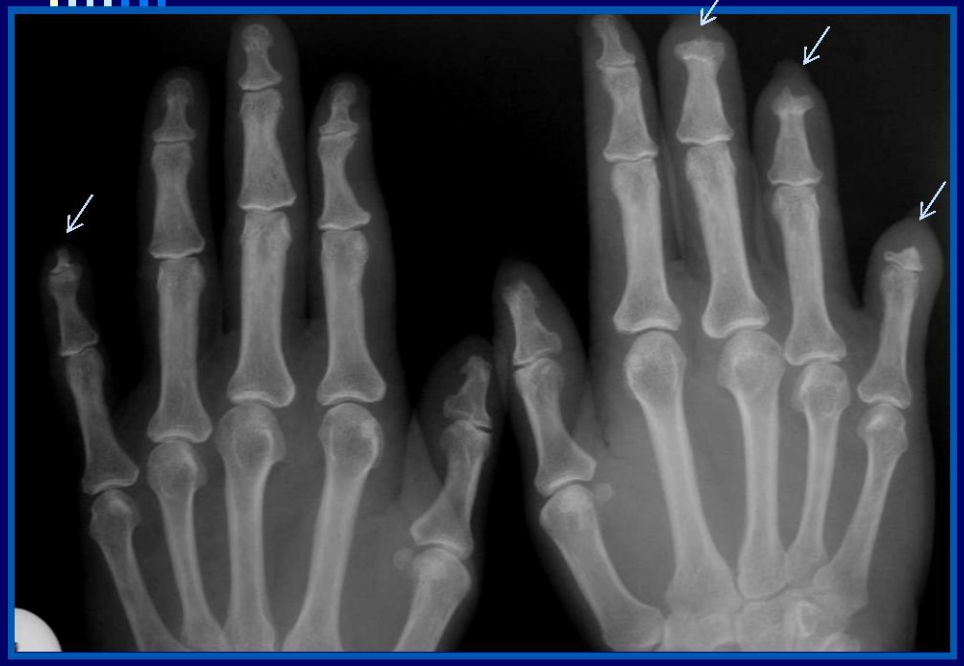
Maladies neurologiques
(arthropathies nerveuses)

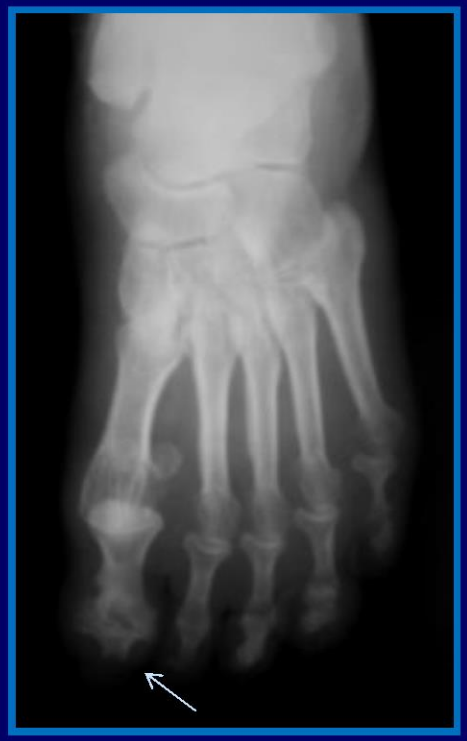
L hyperparathyroïdie primitive/IR dialysés











Patient de 50 ans se plaignant d'acropathies ulcéro-mutilantes depuis 15 ans associées à des troubles de la sensibilité des extrémités.
Radiologiquement on retrouve une acro-ostéolyse longitudinale des mains et des pieds. Il s'agit d'une **arthropathie nerveuse : LÈPRE**

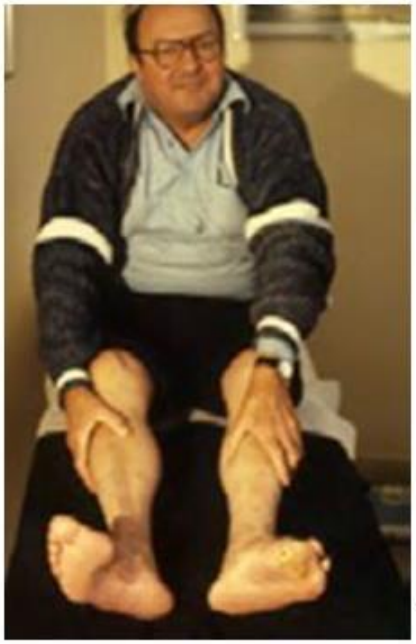
Acropathie ulcéromutilante : la maladie de Thévenard

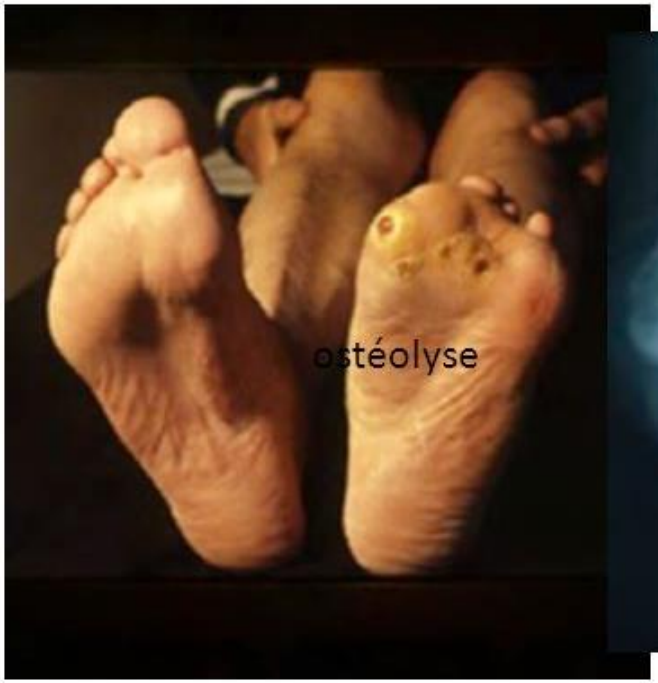
- fille entre 14 et 20 ans+++

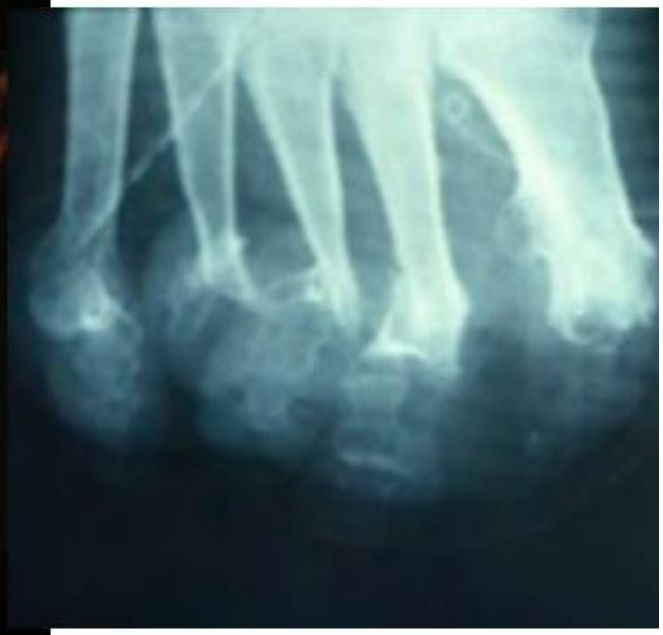
- L'ostéolyse s'étend aux articulations métatarso-phalangiennes voisines puis au tarse, aboutissant au pied cubique.

- L'atteinte des membres supérieurs est rare et toujours associée à celle des membres inférieurs.

Une affection podologique voisine : l'acropathie ulcéro-mutilante





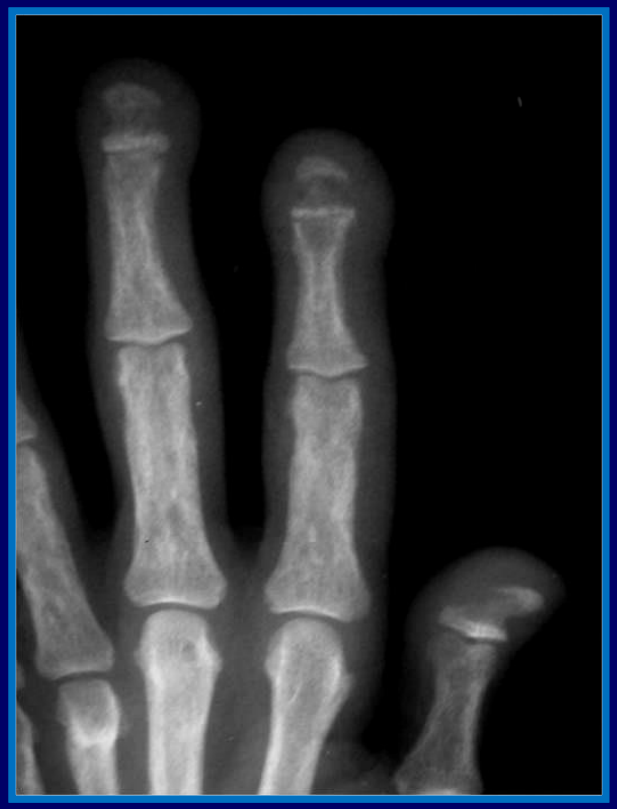


ostéolyse

ATTEINTES METABOLIQUES

Hyperparathyroïdie

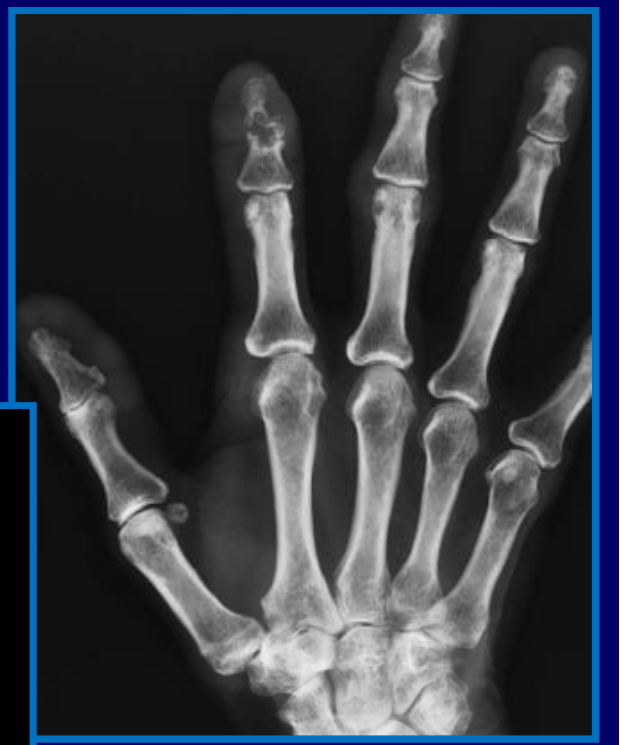
- Primaire ou secondaire
- résorption osseuse sous-périostée du bord radial de la 2ème phalange des 2° et 3° doigts.
- Acro-ostéolyses
- Autres lésions osseuses associées: articulations acromio-claviculaires, crâne, bassin…
- Bilan phospho-calcique perturbé

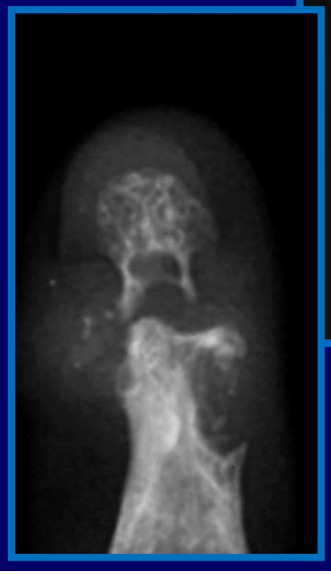


ATTEINTES METABOLIQUES

Arthropathie goutteuse

- Rare actuellement
- Site préférentiel : métatarso-phalangien du 1er rayon
- Signes radiologiques associés: Géodes, Pincement articulaire, Ostéophytes , Tophus (tuméfaction et calcification des parties molles)
- Antécédent d'accès aigue (gros orteil ++)
- Localisations : pieds, mains, poignets, genoux , coudes.
- Hyper-uricémie





SARCOÏDOSE

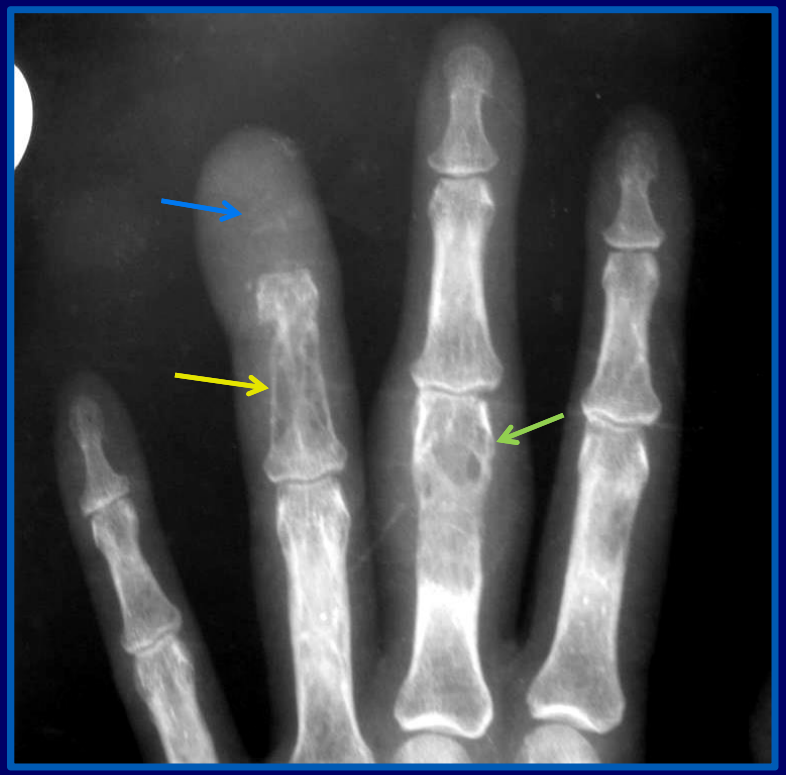
Aspects radiologiques:

Type I : volumineuse géode donnant un aspect évidé à la phalange

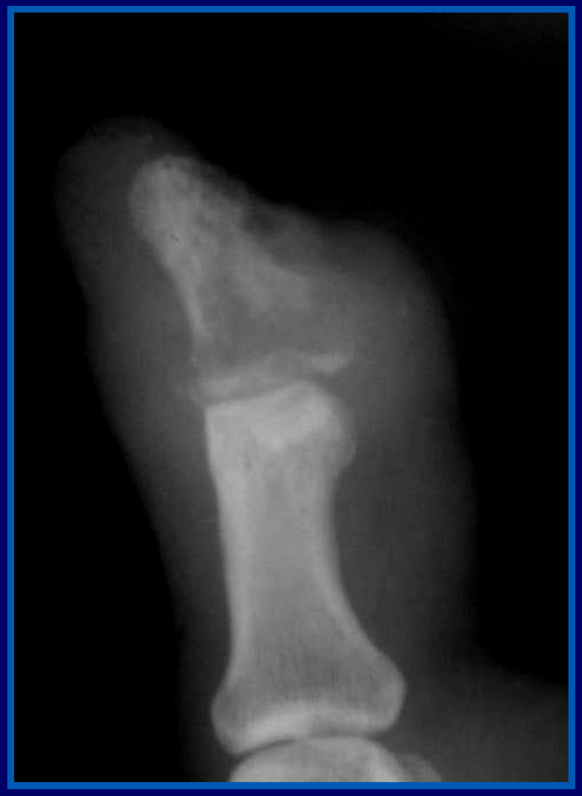
Type II : la tête phalangienne est le siège de multiples petites lacunes pseudo-kystiques (↑)

Type III : forme diffuse microgéodique dite « grillagée », hypertransparence diffuse avec perte de la différentiation cortico-médullaire (↑)

Acro-ostéolyse: forme rare (↑)



INFECTIONS

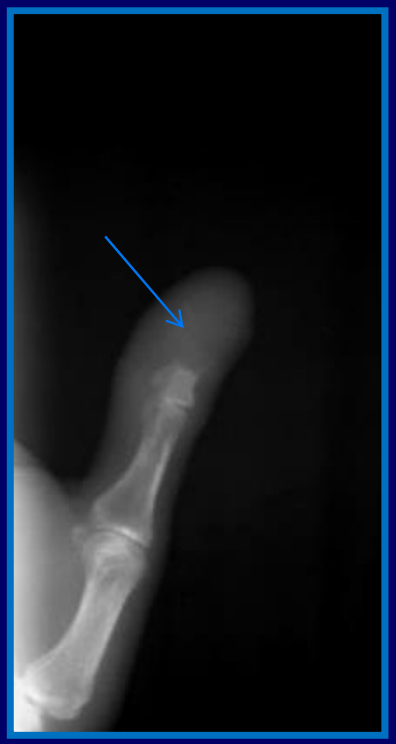


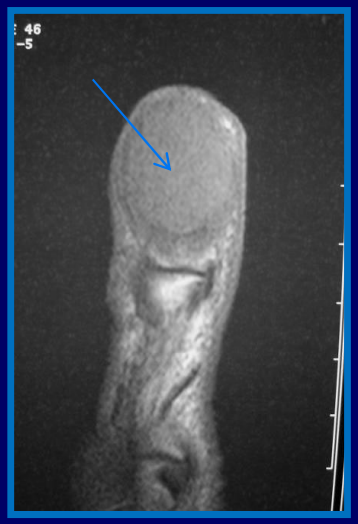
Cas d'une acro-ostéolyse secondaire à un panaris.

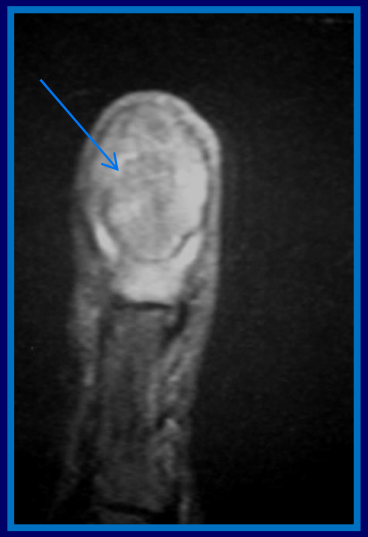
TUMEURS

Acro- métastases

- Rares,
- Souvent révélatrices d'un cancer pulmonaire (50%), mammaire ou digestif.
- Souvent ostéoporose des segments osseux sus ou sous jacents.







Acrométastase d'un carcinome bronchique (Radiographie standard et coupes IRM)

AUTRES CAUSES

Brulures, gelures et traumatismes :

- Une acro-ostéolyse peut être secondaire à une brûlure des doigts (chaleur, électricité) ou à des engelures.
- un stress répété de l'extrémité des doigts, comme chez les joueurs de guitare par exemple, une acro-ostéolyse transversale peut être rencontrée.

Causes toxiques :

- Vapeurs de matières synthétiques : chlorure de vinyl
- Venin de serpent ou de scorpion.
- Médicaments (phénytoïne, ergot de seigle).

Pycnodysostose :

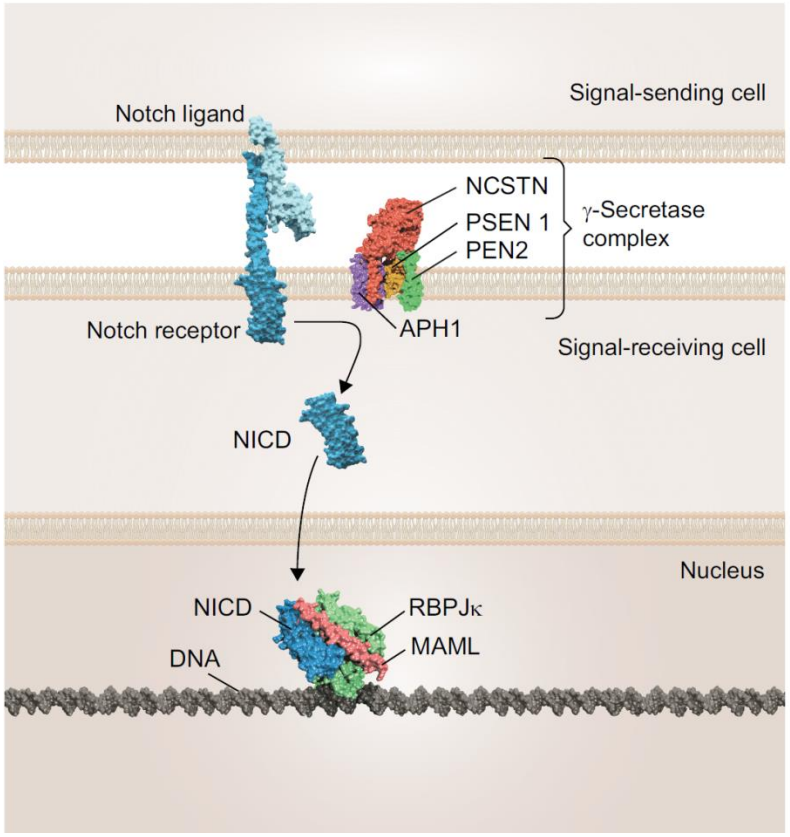
- nanisme avec ostéopétrose et acro-ostéolyse.

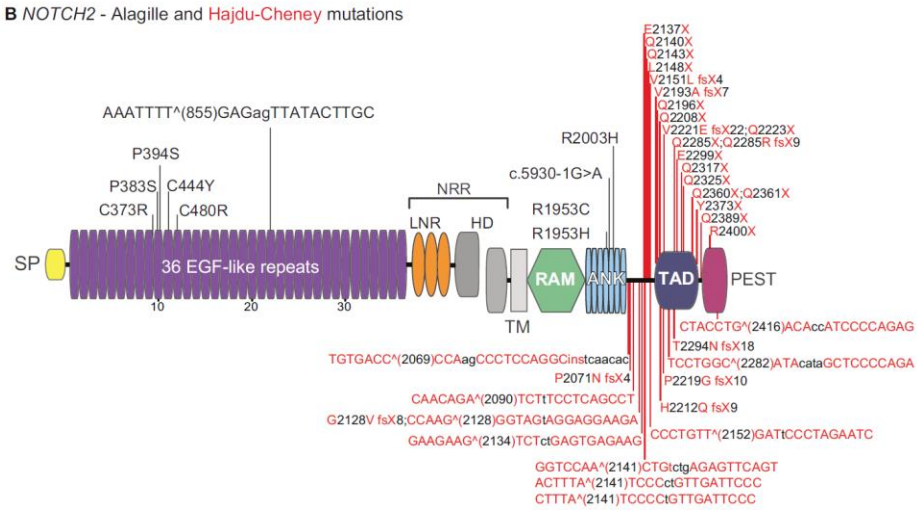
Les acro-ostéolyses génétiques

Syndrome de Hadju et Cheney

Autosomique dominant

mutation de type gain de fonction dans Notch2





Les mutations: siègent toutes dans l'exon 34

Protéine qui n'est plus dégradée dans le protéasome,

Activation permanente de la voie Notch-Hes

THE JOURNAL OF BIOLOGICAL CHEMISTRY VOL. 291, NO. 4, pp. 1538–1551, January 22, 2016

Hajdu Cheney Mouse Mutants Exhibit Osteopenia, Increased Osteoclastogenesis, and Bone Resorption*◆

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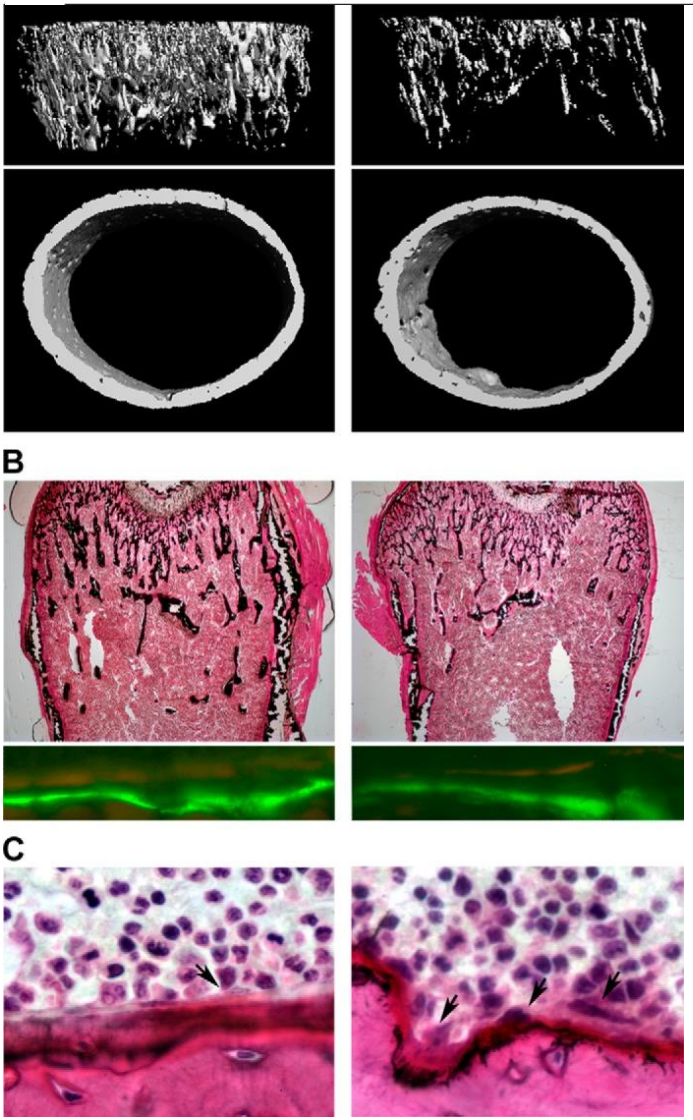
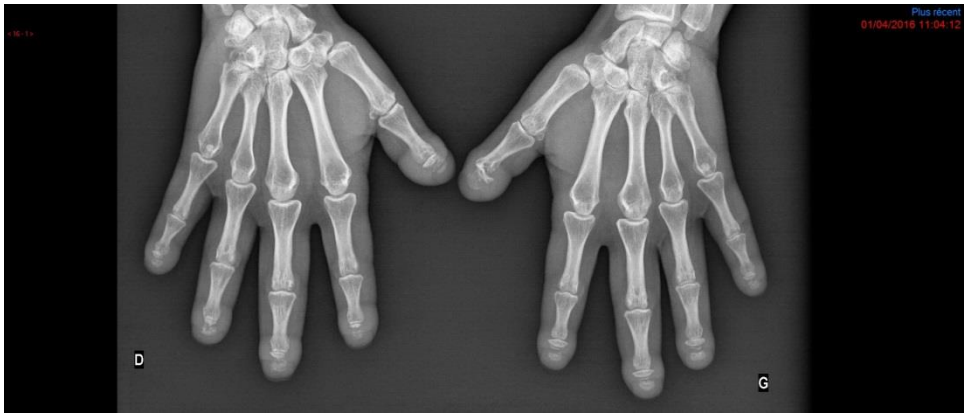


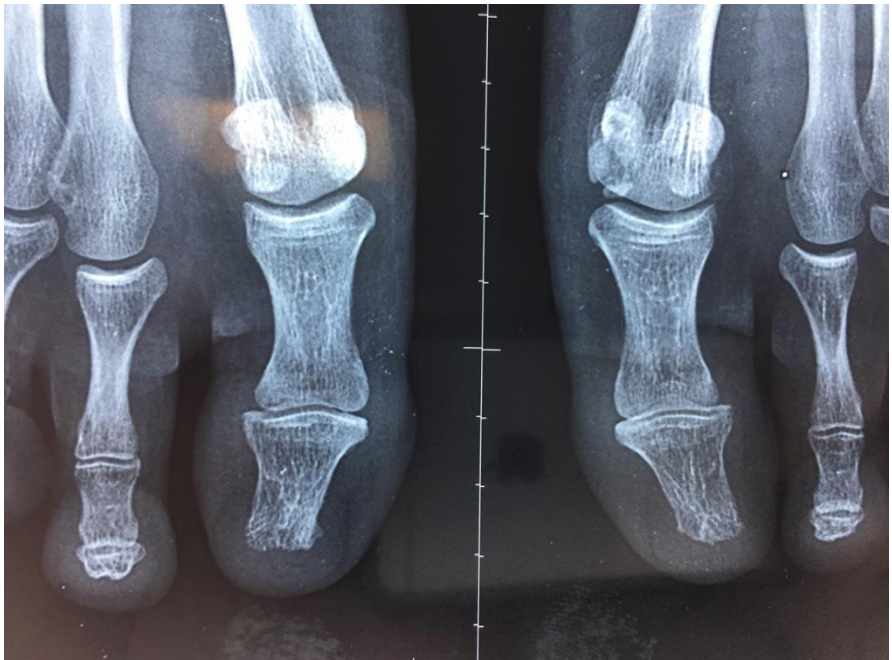
Sur le plan osseux, ces mutations augmentent le recrutement et l'activité des ostéoclastes

Les anomalies phénotypiques des patients affectés concernent essentiellement le squelette et la dentition:

• petite taille,
•dysmorphie faciale avec micrognatisme,
• ostéoporose,
•Cyphoscoliose
• anomalie des os longs
• Edentation précoce par résorption alvéloaire des maxillaires

1 an





Osteoporos Int (2016) 27:1269–1273
DOI 10.1007/s00198-015-3314-8

CASE REPORT

Capillaroscopic findings in a case of Hajdu-Cheney syndrome

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Abstract
Summary Hajdu-Cheney syndrome (HCS) is a rare disease which causes osteoporosis, digit shortening, and early tooth loss. In a young HCS female patient, the nailfold capillaroscopy showed reduced capillary height and reduced density in all affected fingers. Capillaroscopy could improve follow-up and therapy assessment in HCS.

Hajdu-Cheney syndrome (HCS) is a very rare connective tissue disease characterized by osteoporosis, early dentition loss and a particular phenotype as a result of enhanced NOTCH2 clubbing of four fingers and three toes. Hand radiographs showed periarticular osteoporosis and asymmetric bony involvement with acral resorption and/or transversal lucency bands in several fingers. Early collagen-vascular diseases were ruled out by clinical and ancillary examinations, including immunology and immunoblot for systemic sclerosis. Nailfold capillaroscopy showed reduction of capillary height and density in all affected fingers. Notably, in the fingers with acral resorption, many capillaries were dilated, while in the ones with radiolucency band, capillary dilation was a rare

Un patient est suivi dans le service, il souffre surtout de douleurs des doigts secondaire à cette acro ostéolyse.

Un traitement par tildiem a été initié le 9 janvier dans l'hypothèse d'altération de la micro vascularisation. Un dossier similaire est suivi à Cochin, le traitement par tildiem a soulagé le patient.

Les acro-ostéolyses génétiques

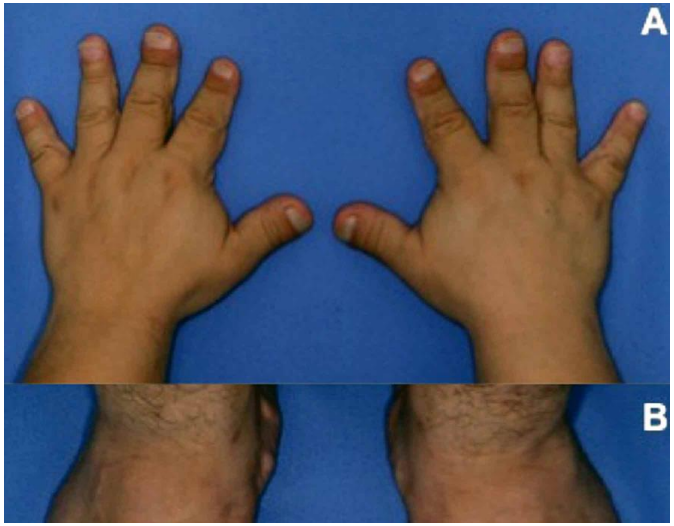
Elles sont exceptionnelles, et ont été rapportées entre autres au cours:

- Des anonichies totales, liée à une mutations dans le gène R-spondin 4 (RSPO4)
- De la pycnodysostose (mutation dans le gène de la Cathepsine K)
- De la dysplasie métaphysaire avec ostéosclérose (mutation dans le gène leucine-rich repeat kinase 1)
- De certaines laminopathies (mutation dans le gène de la lamine) (24).

parents, maternal and neonatal history was uneventful.

The patient reported several episodes of fractures: two of the right tibia, six of the left tibia, one of the left humerus and one of the nasal bones. On physical examination he had the following features: standing height 1.60m; hypermobile extremities; brachydactyly of hands and feet with dysplastic nails (Fig. 1A-B); absence of closing of the fontanels; frontal and occipital bossing (Fig. 2A); midfacial hypoplasia with proptosed eyes (Fig. 2B); anterior open bite (Fig. 3A); narrow high arched grooved palate (Fig. 3B). After clinical examination, the patient underwent radiographic examination with anteroposterior and lateral skull radiographs and anteroposterior radiographs of both hands. Films of the hands showed aplastic distal phalanges in keeping with acro-osteolysis (Fig. 4). Skull films (Fig. 5A-B) showed wide sutures, hypoplastic maxilla, hipopneumatization of the frontal sinuses and the mandible was underdeveloped with an obtuse mandibular angle, being nearly straight.

The clinical and radiological features exhibited by the patient led to a diagnosis of pycnodysostosis.



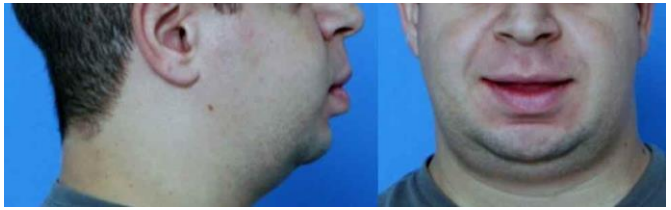


Fig. 2. Lateral (A) and anterior (B) aspects showing frontal and occiptal bossing; hooked nose; midfacial hypoplasia; retrognathia; proptosed eyes and convex facial profile.

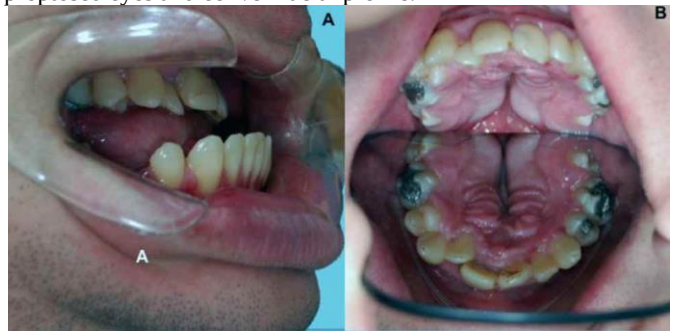


Fig. 3. Anterior open bite (A); narrow high arched grooved palate

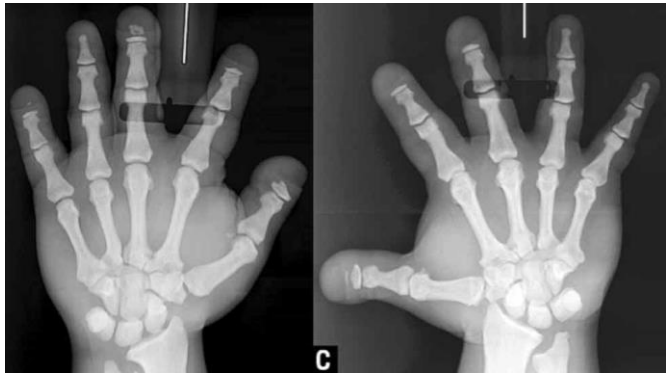
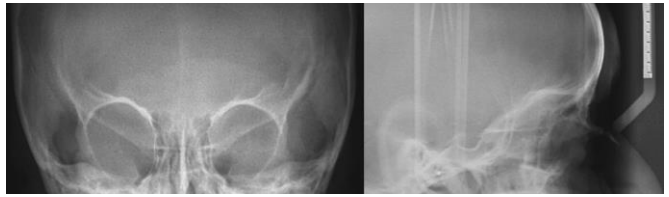


Fig. 4. Hand and wrist radiographs showing the acro-osteolysis of the distal phalanges.



Congenital non-syndromic anonychia totalis with acroosteolysis

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