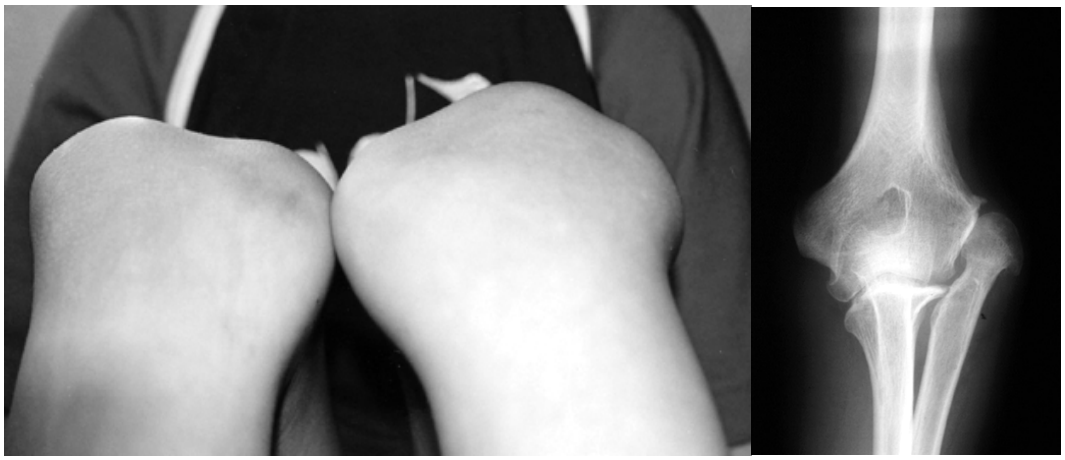
Case Report

30-year-old gentleman, presented with bilateral dislocated radial head and with painful clunk in the knee [bilateral]. He is an active gentleman with good work history. He had a recent soft tissue injury to the neck with rapid deterioration due to severe form of Chronic Regional Pain Syndrome.



Your Diagnosis

Diagnosis: Nail Patella Syndrome

Nail patella syndrome (NPS, hereditary osteo-onychodysplasia (HOOD), Fong disease, Turner-Kieser syndrome, and Österreicher-Turner syndrome] is a pleiotropic condition with a classical clinical tetrad involving the nails, knees, elbows, and the presence of iliac horns.

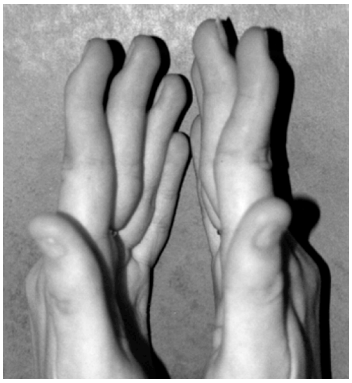
Although the diagnosis may be made at birth, it is very common for families to remain undiagnosed for several generations.

NPS is an autosomal dominant condition.

The incidence: 1 in 50 000

Localisation of the NPS gene: 9q34.

Encoding the transcription factor LMX1B



HAND

1.A sensitive sign of digital involvement in NPS is loss of the creases at the DIP [seen in 96%]

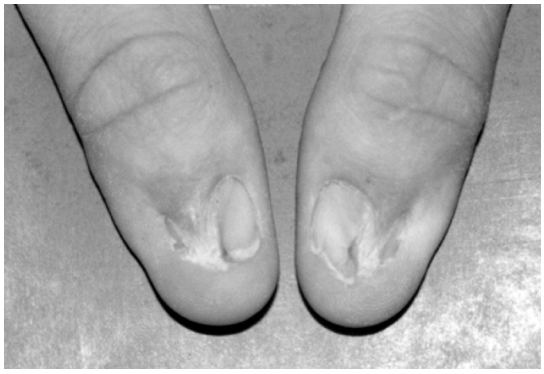
2. Hyperextension of the PIP joint and flexion of the DIP joints, resulting in “swan necking” [58%]

3. Fifth finger clinodactyly was seen in 35%

NAIL AND DIGITAL CHANGES [98%]

Nail changes are the most constant feature of NPS. Nails may be absent, hypoplastic, or dystrophic, ridged longitudinally or horizontally, pitted, discoloured, separated into two halves by a longitudinal cleft or ridge of skin, thin or, less often, thickened. Nail changes may be observed at birth and are most often bilateral and symmetrical.

The thumbnails are the most severely affected, and the severity tends to decrease towards the little finger.



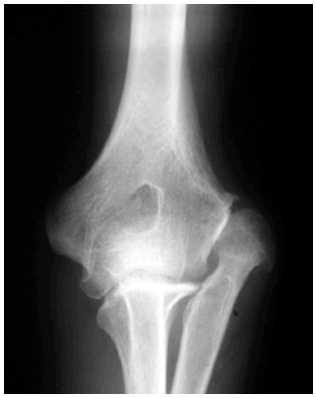
BILATERAL CHRONIC DISLOCATION OF THE RADIAL HEAD:

Usually symmetrical but may be asymmetrical.

Typical radiological findings include dysplasia of the radial head, hypoplasia of the lateral epicondyle and capitellum and prominence of the medial epicondyle.

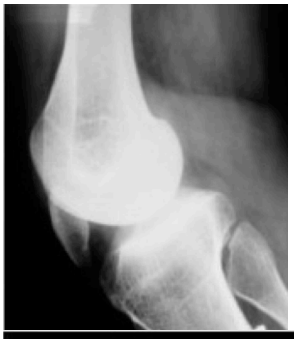
In 33% may be painful.

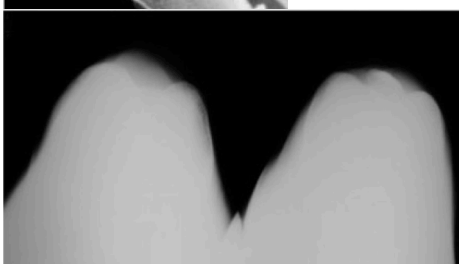
In 12% patient may have pterygium is present.



KNEE

1. The patellae may be small, irregularly shaped
2. Recurrent subluxation of the patella is due to poor development of the vastus medialis muscle.
3. There may be prominent medial femoral condyles, hypoplastic lateral femoral condyles;
4. Early degenerative arthritis is not uncommon.





Symptoms: pain, giving way, a feeling of instability, locking, clicking, patella dislocation, and the inability to straighten the knee joint.

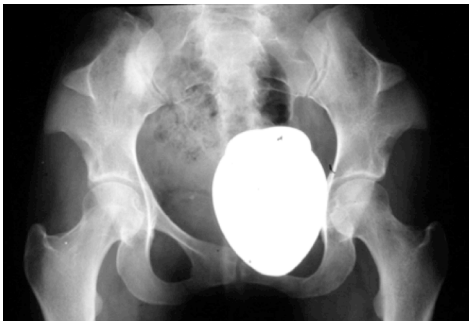
Hypoplastic in 75% ;Patella dislocations 25%

ILIAC HORNS

Iliac horns are bilateral, conical, bony processes that project posteriorly and laterally from the central part of the iliac bones of the pelvis

Large horns may be palpable

Iliac horns were present in 68%



SPINE

Back pain was a problem for 55% of patients.

This was subjectively assessed as moderate or severe in over half of the patients.

Back pain could start during childhood and was the reason that several people had been registered disabled.

An increased lumbar lordosis was seen in 47.1%

GENERALISED PAIN SYNDROME

Some patients described generalised muscular pains, usually worse in cold, damp weather.

Specific diagnoses that had been made included fibromyalgia, polymyalgia rheumatica, and fibrositis.

Two patients had been diagnosed with chronic fatigue syndrome or myeloencephalomyalgia.

RENAL INVOLVEMENT

It is the renal manifestations of NPS that influence mortality.

The main pathology involves a defect in the glomerular basement membrane. The first sign of renal involvement is usually proteinuria, with or without haematuria.

Proteinuria may present at any age from birth onwards and may be intermittent.

May progress to nephrotic syndrome or nephritis, and occasionally to renal failure.

They include irregular thickening of the glomerular basement membrane with electron lucent areas giving a mottled “moth eaten” appearance, and the presence of collagen-like fibres within the basement membrane and the mesangial matrix.

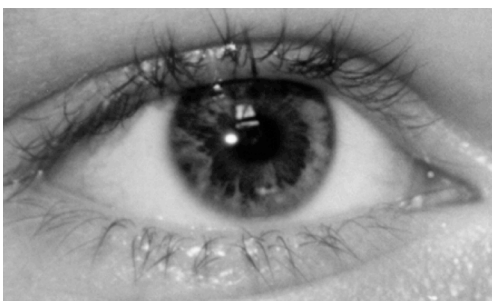
Renal involvement occurred in 25% of patients overall

OPHTHALMOLOGICAL FINDINGS

Glaucoma has been recognised as a feature of NPS.

Primary open angle glaucoma is the most frequent abnormality and the incidence is 9.6%

The pathogenesis of the ocular hypertension and glaucoma in not known.



Lester's sign consists of a zone of darker pigmentation around the central part of the iris, which is roughly a cloverleaf or flower shape. This is most pronounced in blue eyes. Lester's sign was observed in 54%.

NEUROLOGICAL PROBLEMS

Neurological problems have not previously been reported as part of the phenotypic spectrum of NPS.

Several patients spontaneously reported peripheral neurological symptoms and therefore other patients were subsequently asked about these symptoms, which were reported in 25%.

A pattern consisting of intermittent episodes of numbness and tingling and sometimes burning sensations in the hands and sometimes the feet, with no obvious precipitant. The symptoms did not follow the distribution of any particular dermatome or peripheral nerve, the distribution being more like a glove and stocking pattern. These symptoms could spread to the elbows and calves and could last from minutes to hours. There was no history of associated motor weakness.

Epilepsy was reported in 6%

There is a growing interest in the role of LMX1B in the developing brain. Lmx1b is already known to have an important role in the development of mesencephalic dopamine producing cells in mice, is highly expressed in the substantia nigra and ventral tegmental areas, and expression is present from an early stage in development and is maintained throughout life

There are two possible reasons for the increased incidence of peripheral circulatory problems seen in NPS. Firstly, there may be a defect in neuronal migration affecting the digital cutaneous neurones. Secondly, there may be a confounding connective tissue problem in NPS that may predispose to the development of peripheral circulatory problems and Raynaud's phenomenon.

REFERENCE

1. J Med Genet 2003;40:153-162 doi:10.1136/jmg.40.3.153
2. Acta ortop. bras. vol.15 no.4 São Paulo 2007; 141