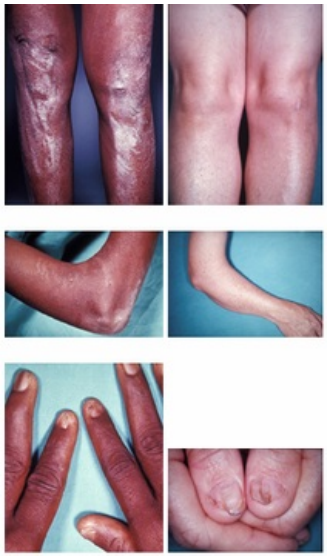
Hereditary osteoonychodysplasia

Synonyms: ***nail-patella syndrome***, HOOD

Definition, Clinical Manifestation, X-ray Imaging

- rare autosomal dominant inherited disease (mutation of the *LMX1B* gene on chromosome 9; role in morphogenesis of the glomerular basement membranes);
- severe deformities (dystrophic changes) of the **nails** or their aplasia (most visible on the thumbs);
- aplasia or hypoplasia of the **patella**;
- dysplastic changes of the elbow (**cubitus valgus**) and knee joint (**genua valga**) – often leading to dislocation of capitulum radii (and restriction of elbow movement) or dislocation of the patella;
- outgrowth at iliacs („**iliac horns**“) + open buckets hipbone with prominent anterior superior iliac spine (image of "**elephant's ear**");
- spondylolisthesis, scoliosis;
- pes equinovaris, congenital flat foot, abnormal iris pigmentation, in 3rd to 4th-decade nephropathy with renal failure can occur (about 1/3 of the patients; preceded by proteinuria and hematuria).



The appearance of a patient with osteo-onychodysplasia.

Nail-patella Syndrome (hereditary osteo-onychodysplasia).

Treatment

- Symptomatic (correction of leg deformities, then possibly patellar dislocation.)

References

Literature

- DUNGL, P.. *Ortopedie.* 1. edition. Praha : Grada Publishing, 2005. ISBN 80-247-0550-8.
- KLENER, P. *Vnitřní lékařství.* 3. edition. Praha : Galén, 2006. ISBN 80-7262-430-X.