

Hereditary osteo-onychodysplasia

synonym: nail-patella syndrome, HOOD

Definition, clinical and x-ray picture

- Rare autosomal dominant hereditary disease (mutation gene in *LMX1B* on the 9th chromosome; role, among other things, in the morphogenesis of the glomerular basement membranes);
- severe deformities (dystrophic changes) of the **nails** or their aplasia (most pronounced on the thumbs);
- aplasia or hypoplasia **patellae**;
- dysplastic changes of the skeleton elbow (**cubitus valgus**) and knee joint (**genua valga**) - often leading to dislocation of the head radius (and limitation of movement in the elbow) or dislocation of the patella;
- outgrowths of the iliac blades ("**iliac horns**") + open iliac blades with protruding spina iliaca anterior superior (image of an "**elephant ear**");
- spondylolisthesis, scoliosis;
- pes equinovarus, congenital flat foot, abnormal pigmentation of the iris, in the 3rd-4th decade of nephropathy with renal failure (in about 1/3 of patients; preceded by proteinuria and hematuria).

Nail-patella syndrome (hereditary osteoonychodysplasia).

Therapy

- Symptomatic (correction of leg deformities, then possible dislocation of the patella).

Links

References

- DUNGL, P., et al. *Orthopedics*. 1. edition. Prague : Grada Publishing, 2005. ISBN 80-247-0550-8.
- KLENER, P, et al. *Internal medicine*. 3. edition. Prague : Galen, 2006. ISBN 80-7262-430-X.



Appearance of a patient with osteonychodysplasia