Nephronophthisis

Nephrophthisis - NPH is a kidney disease of children and young people that leads to kidney failure and accounts for up to 15% of chronic kidney disease in children and adolescents.

Etiopathogenesis

It is a group of rare genetically heterogeneous diseases with **AR** inheritance caused by mutations in NPH1–4 genes - so-called nephrocystins.

Types

- NPH1 and 4 is a juvenile form - other organs are also affected.
- NPH2 in children.
- NPH3 in adolescents.

Clinical picture

- It is characterized by formation of cysts in the medullary or corticomedullary area and by fibrosis.
- The disease first manifests itself (until about 4 years) with polyuria and polydipsia, impaired concentration of kidneys with large losses of salt in urine, weakness, symptoms of developing renal failure. With regard to salt loss, there is not always hypertension.
- Later anemia due to significantly reduced levels of erythropoietin and development of renal failure.

Senior-Loken syndrome - a combination of nephronophthesis with retinitis pigmentosa with early blindness.

Diagnostics

- Genetic.
- Sonographic - only in the stage of chronic renal insufficiency can cystic changes at the interface of the cortex and marrow of normally large or reduced kidneys be demonstrated.
- Histology - in renal biopsy, sclerosing tubulointerstitial nephritis with the development of cyst in the kidney marrow can be demonstrated.
    - Marrow cystic kidney disease has a histological finding indistinguishable from nephronophthisis.

Therapy

- Symptomatic - treatment of kidney failure manifestations including hemodialysis or transplantation.

Links

Related articles

- Polycystic kidney disease/autosomally dominant
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- Hypertension
- Antihypertensives