Nephronophthisis is an autosomal recessive cystic kidney disease. The most frequent genetic cause present at the end-stage renal disease which is up to the third decade of life[1]. These are caused mainly by mutations in 11 different types of genes, and are denoted as nephrocystins (NPHP1-11, NPHP1L)[2]. The increasing numbers of these genes are identified. Recent studies have described that the ciliary expression of the nephrocystins together with the other cystoproteins, such as the polycystins 1 and 2 and the fibrocystin[3]. These findings have shifted our focus to a pathomechanism that involves the defects in the ciliary function the and planar cell polarity[4]. In addition to it many findings regarding the new nephrocystin genes have shown that the disease spectrum of the NPHP is much broader than the previously anticipated one. There are different forms of mutations within the same NPHP gene that can cause various different disease severities [5]. The clinical spectrum has now become even more complex with the possibility of the oligogenicity in the NPHP.

Nephronophthisis is literally means the ‘disappearance of nephrons’. Typical ultrasound features are including in the normal or in the reduced renal size, the loss of cortico-medullary differentiation and the corticomedullary cysts. The Renal biopsy findings include the tubular atrophy, the interstitial fibrosis and the tubular basement membrane defects, including the abrupt transition between the thickening and attenuation or disintegration.

Incidence

This is a rare congenital renal anomaly in which medical council recorded that approximately about 10 to15% of Nephronophthisis patients have extra renal symptoms are found and which[8] includes the retinal degeneration of cerebellar vermis aplasia, liver fibrosis, oculomotor apraxia and also cone-shaped[9].

Ontogenesis of the normal development of the Kidney

The normal embryological development of the kidney is mentioned below with further embryological aspects.[10] There are mainly three kidney systems which are formed in a cranial-to-caudal sequence [11] during the intrauterine life in humans; they are (i) Pronephros (ii) Mesonephros and (iii) Metanephros respectively.

The first of these systems is mainly rudimentary and nonfunctional,[12] but in the second may function for a short time during the early fetal period, and the third forms the permanent kidney.

Fig 1. Normal development of kidney.
the cervical region. [14]These groups form the vestigial excretory units, nephrotomes that regress before it more caudal ones are formed[15]. By the end of this fourth week, the pronephric system will disappear[16].

**Mesonephros**

The Mesonephros and [17] the mesonephric ducts are derived from the intermediate mesoderm from the upper thoracic to the upper lumbar (L3) segments [18]. In the early 4th week of the development, the first excretory tubules of the mesonephros appear[19]. They lengthen rapidly, and forms an S-shaped loop,[20] and which acquire a tuft of the capillaries that will form a glomerulus at[21] their medial extremity. Around the glomerulus, the tubules form Bowman’s capsule,[22] and together these structures constitute into a renal corpuscle.

Laterally, [23] this tubule enters into the longitudinal collecting duct known as the mesonephric or Wolffian duct. [24] While the caudal tubules are still differentiating, the cranial tubules and the glomeruli show the degenerative changes, and by the end of this[25] 2nd month the majority will be disappeared.[26] In the male a few of the[27] caudal tubules and the mesonephric duct persist and participate in formation[28] of the genital system,[29] but they disappear in the female.

**Metanephros**

The third urinary organ[30], the metanephros or permanent kidney appears in the 5th week. [31] The excretory units develop from metanephric mesoderm (blastema) in the same manner as in the mesonephric system. [32] The development of the duct system usually differs from that of the other kidney systems[33].

**Ontogenesis for the Nephronophthisis:**

The abnormal condition [34] is due to the disappearance of the[35] nephrons during the embryonic period [36]. Nephronophthisis that affects the development of the microscopic tubules deep within the kidneys in which that concentrate the[37] urine and which reabsorb the sodium[38]. As a result the excessive amounts of sodium they are excreted in the urine and resulting in too little sodium in the blood and in the body[39]. Excessive amounts of acids may also leads to accumulate in the blood.[40] The damaged tubules become inflamed and scarred,[41] eventually these causing leads to the chronic kidney disease severe enough to[42] result in end-stage renal disease.

**Discussion**

Nephronophthisis is literally means the ‘disappearance of nephrons’[43]. Typical ultrasound features are including in the normal or in the reduced renal size, the loss of cortico medullary differentiation and the cortico medullary cysts.[44] The Renal biopsy findings include [45] the tubular atrophy, the interstitial fibrosis and the tubular basement membrane defects, including the abrupt transition between the[46] thickening and attenuation or disintegration. [47] There are different forms of mutations within the same NPHP gene [48] that can cause various different disease severities. The clinical spectrum has[49] now become even more complex with the possibility of the oligogenicity in the[50] NPHP. There are various types of Nephronophthisis like [51] Juvenile Nephronophthisis, Adolescent Nephronophthisis and Infantile Nephronophthisis [52].

Chronic kidney disease[53] which causes many problems throughout the body. When the loss of kidney function is mild or moderately severe, the kidneys cannot absorb the water from [54] the urine to reduce the volume of the urine and concentrate it. The[55] kidneys have less ability to excrete the acids normally and is produced by the body and the[56] blood becomes more acidic, and this condition is called acidosis. The production of the red blood cells [57] decreases and leads to anemia.[58] High level of the metabolic waste products in the blood can damage the[59] nerve cells in the brain, trunk, arms, and the legs. The Uric acid levels may increase but sometimes causing gout. This diseased kidneys produce hormones that increase the blood pressure. [60] In addition to it the diseased kidneys cannot excrete excess of salt and water. This salt and water retention can contribute to high blood pressure and a chance of heart failure. [61] Through Blood tests, urine tests, Ultrasonography & Sometimes biopsy also we can diagnose the defect.

In normal development [62] There are mainly three kidney systems which are formed in a cranial-to-caudal sequence[63] during the intrauterine life in humans. The first of these systems is mainly rudimentary and nonfunctional,[64] but in the second may function for a short time during the early fetal
period\cite{65}, and the third forms \cite{66} the permanent kidney. Certain treatment measures are\cite{67} the restricting protein, controlling acidosis,\cite{68},\cite{69} lowering triglyceride levels, restricting sodium and potassium and controlling the phosphorus levels\cite{70}.

**Conclusion**

Nephronphthisis is an autosomal recessive cystic kidney disease. From the practical point of view the diagnosis of the NPH should be considered if a child presents with polyuria, urinary sodium loss, growth failure, renal insufficiency without hematuria or proteinuria the normal blood pressure, and the normal-sized kidneys without the dilatation of the urinary tract. These patients should be administrated for screening for homozygous or heterozygous NPHP1 deletion, which are found in 20 to 40% of this type cases. In the absence of heterozygous NPHP1 deletion, the renal biopsy may be proposed to confirm the diagnostic process. At present the screening for mutation in all the other NPHP genes is not performed well due to the low frequency in the detected mutations and the high cost of the procedure.

**References**


