Benign Familial Hematuria

Alternative Names
BFH
Thin-Basement-Membrane Nephropathy
Thin Membrane Nephropathy
TMN

WHO International Classification of Diseases
Diseases of the genitourinary system

OMIM Number
141200

Mode of Inheritance
Autosomal dominant

Description
Benign familial hematuria, also known as thin-basement-membrane nephropathy, is a nonprogressive inherited kidney disorder that usually begins during childhood. It is characterized by the presence of persistent or recurrent hematuria (red blood cells in the urine). Diffuse attenuation of the glomerular basement membrane is usually considered the hallmark of the condition, but it is not specific. The pattern of inheritance of familial benign hematuria is autosomal dominant, and the prognosis of the condition is excellent. However, the diagnosis of this benign disease may be difficult to establish because it is based on a series of negative findings, such as absence of proteinuria, renal failure, or extra-renal symptoms, and the finding of a nonspecific ultrastructural lesion, the thin glomerular basement membrane, and above all on the results of family investigations demonstrating the absence of progression toward renal failure.

Molecular Genetics
Some cases of benign familial hematuria are due to heterozygosity for a mutation in codon 897 of the COL4A4 gene resulting in substitution of a glutamic acid residue for glycine. Missense mutations in the COL4A3 gene can also cause familial benign hematuria. A mutation in exon 36 of the COL4A3 gene that resulted in a gly1015-to-glu (G1015E) amino acid substitution in the collagenous domain of the protein has been identified.

Both the COL4A4 and COL4A3 genes each encode one of the six subunits of type IV collagen, the major structural component of basement membranes. The genes are organized in a head-to-head conformation with other type IV collagen genes so that each gene pair shares a common promoter.

Epidemiology in the Arab World

United Arab Emirates
Abou-Chaaban et al. (1997) studied the pattern of pediatric renal diseases among children in the Dubai Emirate during the period from 1991 to 1996. In this period, a total of 712 pediatric patients, including 230 nationals of the United Arab Emirates, were seen with various renal problems. In their study, Abou-Chaaban et al. (1997) observed six cases of hematuria among nationals of the United Arab Emirates. Interestingly, the male: female ratio in this group was approximately 2:1.

References

Contributors
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