Papillorenal Syndrome

**Alternative Names**
Renal-Coloboma Syndrome
Optic Nerve Coloboma with Renal Disease
Coloboma of Optic Nerve with Renal Disease
Optic Coloboma, Vesicoureteral Reflux, and Renal Anomalies

**WHO International Classification of Diseases**
Congenital malformations, deformations and chromosomal abnormalities

**OMIM Number**
120330

**Mode of Inheritance**
Autosomal dominant

**Gene Map Locus**
10q24.3-q25.1

**Description**
Papillorenal syndrome is a very rare developmental disorder involving optic nerve colobomas and renal hypoplasia/insufficiency, which exhibits autosomal dominant inheritance and a highly variable phenotype even within members of affected families.

**Molecular Genetics**
Papillorenal syndrome is caused by mutations in the PAX2 gene. PAX2 encodes paired box gene 2, one of many human homologues of the Drosophila melanogaster gene prd. The central feature of this transcription factor gene family is the conserved DNA-binding paired box domain. PAX2 is believed to be a target of transcriptional suppression by the tumor suppressor gene WT1. Alternative splicing of this gene results in multiple transcript variants.

**Epidemiology in the Arab World**

**Oman**
Al-Gazali et al. (2000) reported four cases from an extended and highly inbred Arab family from Oman with autosomal dominant inheritance of a syndrome characterized by a variable combination of optic nerve colobomas, renal abnormalities, vesicoureteral reflux, lax joints and arthrogryposis multiplex. The four studied cases represented the 27-year-old father and three male children. Apart from the arthrogryposis multiplex which has not been described in the papillorenal syndrome, the features of the syndrome in this family are very similar to the papillorenal syndrome. However, sequencing of all 12 exons of PAX2 gene revealed no mutation in this family. Hence, the disorder in this family is likely to represent a new syndrome with features overlapping with the papillorenal syndrome.

**References**

**Contributors**
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