Atlas of Genetics and Cytogenetics in Oncology and Haematology

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Gene Section
Mini Review

PAX2 (Paired box gene 2)

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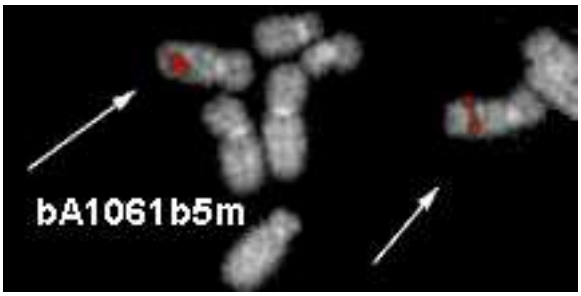
Identity

Other names: Paired box homeotic gene 2

HGNC (Hugo): PAX2

Location: 10q24



Probe(s) - Courtesy Mariano Rocchi, Resources for Molecular Cytogenetics

DNA/RNA

Description

12 exons, including alternative spliced exons 6 and 10.

Transcription

7 alternative splicing isoforms.

Pseudogene

No.

Protein

Description

416 amino acids; 44.7 kDa.

Expression

PAX2 is expressed in the developing eye, ear, central nervous system (CNS), spinal cord, pancreas and urogenital tract. PAX2 contains a DNA binding paired domain, a truncated homeodomain, an octapeptide region and a carboxyl-terminal transactivation domain.

Localisation

Nuclear.

Function

PAX2 is a transcription factor that acts to regulate the expression of genes involved in mediating cell proliferation and growth, resistance to apoptosis, and cell migration. PAX2 null mutant mice die perinatally with absent cochlea, kidneys, ureters, oviducts, vas deferens and epididymis, also demonstrating mid- and hindbrain deficiency and defective optic nerve.

Homology

PAX2 shares homology through the conserved paired box domain with the other members of the nine strong PAX gene family.

Mutations

Germinal

A number of PAX2 mutations have been reported as associated with Renal Coloboma Syndrome (see below), oligomeganephronia and isolated renal hypoplasia. These are collated on the Human PAX2 Allelic Variant Database (see below).

Somatic

See below.

Implicated in

Renal Coloboma Syndrome (RCS)

Note

Caused by heterozygous PAX2 mutations. Increased apoptosis arising as a result of impaired PAX2 function believed to be responsible for disrupted nephron formation and arrested cochlear outgrowth.

Prognosis

Renal Coloboma Syndrome is associated with end-stage renal failure and blindness.

Various cancers, including carcinomas of the kidney, carcinomas of the prostate, carcinomas of the breast and carcinomas of the ovary. Also Wilms' tumour and Kaposi's sarcoma.

Disease

PAX2 is overexpressed in a variety of cancers (see below). A detailed understanding of the mechanistic contribution mediated by PAX2 has yet to be established. However, inhibition of PAX2 expression in a number of cancer cell lines induces cell death, indicating a role for PAX2 in tumour cell survival.

Prognosis

PAX2 is not expressed in normal prostate tissue or benign prostatic hyperplasia, but is expressed in primary prostatic carcinomas suggesting that PAX2 expression may be a useful marker of prostate cancer. PAX2 expression correlates with proliferation index in the majority of kidney tumour subtypes, and expression levels are significantly higher in patients presenting metastatic disease. PAX2 may therefore provide a useful prognostic marker for determining the severity of kidney cancers.

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