Gene Section
Short Communication

CEP57 (centrosomal protein 57kDa)

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Identity

Other names: PIG8, TSP57, Translokin, KIAA0092
HGNC (Hugo): CEP57
Location: 11q21

DNA/RNA

Description
CEP57 spans over 42 kb and is composed of 11 exons.

Protein

Description
500 amino acids, 57 kDa.

Expression
Ubiquitously expressed.

Localisation
Nucleus, cytoplasm, cytoskeleton, centrosome.

Function
Centrosomal protein required for microtubule attachment to centrosomes.
Also involved in intracellular bidirectional trafficking of factors such as FGF2 along microtubules.

Homology
The CEP57 gene is conserved in chimpanzee, dog, cow, mouse, rat, chicken, and zebrafish.

Figure 1. Schematic representation of CEP57 demonstrating the relative exon sizes.
Figure 2. Schematic representation of CEP57 demonstrating significant functional or structural domains.

Figure 3. Schematic representation of CEP57 demonstrating the relative exon sizes and positions of known mutations. Biallelic mutations are represented by coloured lines, with mutations in the same individual in matching colours.

Mutations

Germinal
Biallelic, loss-of-function mutations in CEP57 have been found in three MVA pedigrees (figure 3).

Implicated in
Mosaic variegated aneuploidy syndrome (MVA)

Note
MVA is a rare recessive condition characterised by mosaic aneuploidies, predominantly trisomies and monosomies, involving multiple different chromosomes and tissues. Affected individuals typically present with severe intrauterine growth retardation and microcephaly. Eye anomalies, mild dysmorphism, variable developmental delay and a broad spectrum of additional congenital abnormalities and medical conditions may also occur.

Prognosis
The prognosis for an individual with MVA syndrome is based on the malformations present in the individual. There is early mortality in a significant proportion of cases due to failure to thrive and/or complications of congenital abnormalities, epilepsy, infections or malignancy.

Cytogenetics
MVA is characterised by mosaic aneuploidies, predominantly trisomies and monosomies, involving multiple different chromosomes and tissues. The proportion of aneuploid cells varies but is usually >10% and is substantially greater than in normal individuals. Some patients with MVA also demonstrate premature chromatid separation in colchicine-treated blood lymphocyte and fibroblast cultures.

Oncogenesis
The risk of malignancy in MVA is high with Wilms tumour, rhabdomyosarcoma, leukaemia and granulosa cell tumour of the ovary reported in several cases. Myelodysplastic syndrome has also been observed.

To be noted
Note
Biallelic mutations in BUB1B have also been identified in individuals with MVA syndrome.

References
Lane AH, Aijaz N, Galvin-Parton P, Lanman J, Mangano R, Wilson TA. Mosaic variegated aneuploidy with growth...


This article should be referenced as such: