Centrosomal Protein, 97-kD

Alternative Names
CEP97

Record Category
Gene locus

WHO-ICD
N/A to gene loci

Incidence per 100,000 Live Births
N/A to gene loci

OMIM Number
615864

Mode of Inheritance
N/A to gene loci

Gene Map Locus
3q12.3

Description
The CEP97 gene encodes a cytosolic protein that forms a complex with the CP110 protein. This complex localizes to the centrosome, a microtubule organizing center consisting of two orthogonally arranged centrioles and a pericentriolar matrix. Centrioles are involved in two major cellular processes: cell division and ciliogenesis. During mitosis, centrioles help in the formation of spindle fibers that separate the chromosomes and during the G1 phase of the cell cycle, they help in ciliogenesis by forming basal bodies that nucleate cilia. These cilia are resorbed into the cell before the next mitosis cycle, presumably as they interfere with mitosis. The protein complex of CEP97 and CP110 is believed to help the centrosome transition into its role in mitosis by capping the mother centriole and inhibiting ciliogenesis.

Molecular Genetics
The CEP97 gene is located on the long arm of chromosome 3. It spans a length of 46.6 kb of DNA and its coding sequence is spread across 11 exons. It encodes a 96.9 kDa protein product comprised of 865 amino acids. An additional 99.9 kDa isoform of the CEP97 protein exists due to an alternatively spliced transcript variant. The gene is found to be overexpressed in the heart.

Epidemiology in the Arab World
Saudi Arabia
Monies et al. (2017) investigated the findings of 1000 diagnostic panels and exomes carried out at a next generation sequencing lab in Saudi Arabia. One patient, an 8-year-old female, presented with primordial short stature and growth parameters below the 5th percentile. She suffered from hearing deficit, developmental and speech delay and behavioral disturbances such as ADHD. Skeletal anomalies included thickness in the base of the skull with narrowing of the foramen magnum, small facial bones, kyphosis in the cervical spine and sacrum, coxa valgus, DDH, general osteopenia, and clinodactyly suggestive of Seckel syndrome. Using whole exome sequencing, a heterozygous mutation (c.1737G>A, p.W579X) was identified in exon 11 of the patient's CEP97 gene. As a mutation in the gene had previously been tentatively linked to intellectual disability, solitary kidney, brachydactyly and facial dysmorphism, this finding helped confirm its association with the phenotype.

References
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Related CTGA Records

External Links

http://www.genecards.org/cgi-bin/carddisp.pl?gene=CEP97

Contributors

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13.09.2017