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Gene Section
Short Communication

LMDRA (leucine rich melanocyte differentiation associated)

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Abstract

C10orf11 encodes a leucine-rich repeat protein having a role in melanocyte differentiation. Mutations in this gene have been associated with autosomal recessive oculocutaneous albinism 7 (OCAVII).

Keywords: OCAVII, albinism, C10orf11

Identity

Other names: C10orf11, CDA017

HGNC (Hugo): LRMDA

Location: 10q22.3

DNA/RNA

Description

In Chromosome: 10, the 1,128,715 bases long gene starts from 75,431,453bp from pter and ends 76,560,167 bp from pter;
Orientation: Plus strand. It contains 6 exons.

Transcription

C10orf11 encodes 16 splice variants of which 4 are protein coding and the remaining are processed transcripts.

Protein

Description

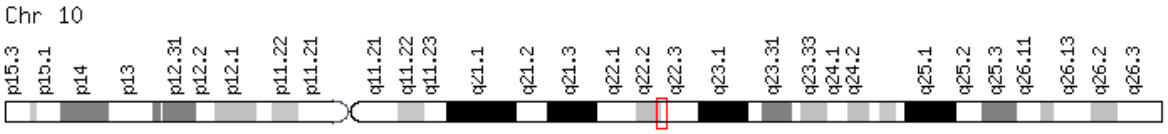
The gene encodes a 198 amino acids long leucine-rich repeat-containing protein of molecular mass 22568 Da.

Expression

The gene is expressed in embryonic melanoblasts and fetal melanocyte and has not been detected in retinal pigment epithelial cells. In addition the expression of the gene in the following tissue types are evident by its existence in the corresponding cDNA libraries: adrenal cortex, brain, cartilage, cerebellum, endocrine, eye, fetus, heart, kidney, liver, lung, muscle, nervous, pancreas, pancreatic islet, placenta, pooled tissue, prostate, skin, stem cell, testis and uterus (http://cgap.nci.nih.gov/Genes/GeneInfo?ORG=Hs CID=118161LLNO=83938).

Localisation

10q22.3



Cytogenetic band showing C10orf11 locus (http://www.genecards.org/cgi-bin/carddisp.pl?gene=C10orf11&keywords= C10orf11).

Function

The precise function of C10ORF11 is not yet known. However, there is some evidence that the protein might have a role in melanocyte differentiation.

Mutations

Germinal

C10orf11 mutations are responsible for Oculocutaneous Albinism type 7 (OCA7). Nine Faroese patients and one Danish patient of Lithuanian origin were found to have mutations in C10orf11 gene representing OCAVII (Gronskov et al., 2014). These patients have a light skin pigmentation that is reported to be lighter than their relatives. Hair color ranges from light blond to dark brown. Eye findings include nystagmus, iris transillumination, visual acuity ranging from 6/9 to 3/60 and very sparse peripheral ocular fundus pigmentation.

Implicated in

Breast Cancer

To identify genetic polymorphisms associated with clinical outcomes of breast cancer patients with tamoxifen treatment, genome-wide association study was conducted using 462 Japanese patients with hormone receptor-positive, invasive breast cancer receiving adjuvant tamoxifen therapy. The
study revealed that rs10509373 in C10orf11 gene to be significantly associated with recurrence-free survival in the replication study (log-rank P= 2.02×10^{-4}).

Hazard ratio per C allele of rs10509373 was found to be 4.51 [95% confidence interval (CI), 2.727.51; P= 6.29×10^{-9}].

In a combined analysis of rs10509373 genotype with previously identified genetic makers, CYP2D6 and ABCC2, the number of risk alleles of these three genes was reported to have cumulative effect on recurrence-free survival among 345 patients receiving tamoxifen monotherapy (log-rank P= 2.28×10^{-12}) (Kiyotani et al., 2011).

References

Grønskov K, Dooley CM, Østergaard E, Kelsh RN, Hansen L, Levesque MP, Vilhelmsen K, Møllgård K, Stemple DL, Rosenberg T. Mutations in c10orf11, a melanocyte-differentiation gene, cause autosomal-recessive albinism. Am J Hum Genet. 2013 Mar 7;92(3):415-21

Kiyotani K, Mushiroda T, Tsunoda T, Morizono T, Hosono N, Kubo M, Tanigawara Y, Imamura CK, Flockhart DA, Aki F, Hirata K, Takatsuka Y, Okazaki M, Ohsumi S, Yamakawa T, Sasa M, Nakamura Y, Zembutsu H. A genome-wide association study identifies locus at 10q22 associated with clinical outcomes of adjuvant tamoxifen therapy for breast cancer patients in Japanese. Hum Mol Genet. 2012 Apr 1;21(7):1665-72

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