SMOC1 gene

SPARC related modular calcium binding 1

Normal Function

The *SMOC1* gene provides instructions for making a protein called secreted modular calcium-binding protein 1 (SMOC-1). This protein is found in basement membranes, which are thin, sheet-like structures that support cells in many tissues and help anchor cells to one another during embryonic development. The SMOC-1 protein attaches (binds) to many different proteins and is thought to regulate molecules called growth factors that stimulate the growth and development of tissues throughout the body. These growth factors play important roles in skeletal formation, normal shaping (patterning) of the limbs, as well as eye formation and development. The SMOC-1 protein also likely promotes the maturation (differentiation) of cells that build bones, called osteoblasts.

Health Conditions Related to Genetic Changes

Ophthalmo-acromelic syndrome

At least 12 mutations in the *SMOC1* gene have been found to cause ophthalmo-acromelic syndrome, a condition that results in malformations of the eyes, hands, and feet. Most of these mutations likely result in a nonfunctional SMOC-1 protein. The loss of SMOC-1 could disrupt growth factor signaling, which would impair the normal development of the skeleton, limbs, and eyes. These changes likely underlie the signs and symptoms of ophthalmo-acromelic syndrome.

Coloboma

MedlinePlus Genetics provides information about Coloboma

Other Names for This Gene

- secreted modular calcium-binding protein 1
- SMOC1_HUMAN
- SPARC-related modular calcium-binding protein 1

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of SMOC1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=64093[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28SMOC1%5BTIAB%5D%29+OR+%28SPARC+related+modular+calcium+binding+1%5BTIAB%5D%29%29+OR+%28secreted+modular+calcium-binding+protein+1%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- SPARC-RELATED MODULAR CALCIUM-BINDING PROTEIN 1; SMOC1 (https://omim.org/entry/608488)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/64093)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=SMOC1[gene])

References

- Abouzeid H, Boisset G, Favez T, Youssef M, Marzouk I, Shakankiry N, Bayoumi N, Descombes P, Agosti C, Munier FL, Schorderet DF. Mutations in the SPARC-relatedmodular calcium-binding protein 1 gene, SMOC1, cause waardenburg anophthalmiasyndrome. Am J Hum Genet. 2011 Jan 7;88(1):92-8. doi: 10.1016/j.ajhg.2010.12.002.Epub 2010 Dec 30. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/21194680) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3014360/)
- Choi YA, Lim J, Kim KM, Acharya B, Cho JY, Bae YC, Shin HI, Kim SY, Park EK. Secretome analysis of human BMSCs and identification of SMOC1 as an important ECMprotein in osteoblast differentiation. J Proteome Res. 2010 Jun 4;9(6):2946-56. doi: 10.1021/pr901110q. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/20359165)
- Klemencic M, Novinec M, Maier S, Hartmann U, Lenarcic B. The heparin-bindingactivity of secreted modular calcium-binding protein 1 (SMOC-1) modulates itscell adhesion properties. PLoS One. 2013;8(2):e56839. doi:10.1371/journal.pone.0056839. Epub 2013 Feb 21. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/23437253) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/arti

cles/PMC3578922/)

- Okada I, Hamanoue H, Terada K, Tohma T, Megarbane A, Chouery E, Abou-Ghoch J,Jalkh N, Cogulu O, Ozkinay F, Horie K, Takeda J, Furuichi T, Ikegawa S, NishiyamaK, Miyatake S, Nishimura A, Mizuguchi T, Niikawa N, Hirahara F, Kaname T,Yoshiura K, Tsurusaki Y, Doi H, Miyake N, Furukawa T, Matsumoto N, Saitsu H. SMOC1 is essential for ocular and limb development in humans and mice. Am J HumGenet. 2011 Jan 7;88(1):30-41. doi: 10.1016/j.ajhg.2010.11.012. Epub 2010 Dec 30. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/21194678) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3014372/)

- Rainger J, van Beusekom E, Ramsay JK, McKie L, Al-Gazali L, Pallotta R,Saponari A, Branney P, Fisher M, Morrison H, Bicknell L, Gautier P, Perry P,Sokhi K, Sexton D, Bardakjian TM, Schneider AS, Elcioglu N, Ozkinay F, Koenig R,Megarbane A, Semerci CN, Khan A, Zafar S, Hennekam R, Sousa SB, Ramos L,Garavelli L, Furga AS, Wischmeijer A, Jackson IJ, Gillessen-Kaesbach G, BrunnerHG, Wieczorek D, van Bokhoven H, Fitzpatrick DR. Loss of the BMP antagonist,SMOC-1, causes Ophthalmo-acromelic (Waardenburg Anophthalmia) syndrome in humansand mice. PLoS Genet. 2011 Jul;7(7):e1002114. doi: 10.1371/journal.pgen.1002114.Epub 2011 Jul 7. Erratum In: PLoS Genet. 2018 Dec 26;14(12):e1007866. doi:10.1371/journal.pgen.1007866. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/21750680) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3131273/)

Genomic Location

The *SMOC1* gene is found on chromosome 14 (https://medlineplus.gov/genetics/chromosome/14/).

Last updated March 1, 2014