ESCO2 gene

establishment of sister chromatid cohesion N-acetyltransferase 2

Normal Function

The *ESCO2* gene provides instructions for making a protein that is important for proper chromosome separation during cell division. Before cells divide, they must copy all of their chromosomes. The copied DNA from each chromosome is arranged into two identical structures, called sister chromatids. The ESCO2 protein plays an important role in establishing the glue that holds the sister chromatids together until the chromosomes are ready to separate.

Health Conditions Related to Genetic Changes

Roberts syndrome

At least 30 *ESCO2* gene mutations have been found to cause Roberts syndrome, which is characterized by limb and facial abnormalities and slow growth before and after birth. These mutations prevent the cell from producing any functional ESCO2 protein. Some mutations change single protein building blocks (amino acids), while others result in an abnormally short protein. The absence of functional ESCO2 protein causes some of the glue between sister chromatids to be missing around the chromosome's constriction point (centromere). In Roberts syndrome, cells respond to abnormal sister chromatid attachment by delaying cell division. Delayed cell division can be a signal that the cell should undergo self-destruction. The signs and symptoms of Roberts syndrome may be due to the loss of cells from various tissues during early development.

Researchers originally suspected that the varying severity of Roberts syndrome was caused by different types of mutations in the *ESCO2* gene. They predicted that people with the mild form of the disorder would have mutations that reduced the activity of the ESCO2 protein, while those with the severe form would have mutations that completely eliminated the protein's function. However, all known mutations in the *ESCO2* gene prevent the production of any functional ESCO2 protein. The underlying cause of the variation in disease severity remains unknown. Researchers suspect that other genetic and environmental factors may be involved.

Other Names for This Gene

- EFO2

- ESCO2_HUMAN
- establishment of cohesion 1 homolog 2
- establishment of cohesion 1 homolog 2 (S. cerevisiae)

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of ESCO2 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=157570[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28ESCO2%5BTIAB%5D%29+OR+%28EFO2%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- ESTABLISHMENT OF SISTER CHROMATID COHESION N-ACETYLTRANSFERASE 2; ESCO2 (https://omim.org/entry/609353)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/157570)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=ESCO2[gene])

References

- Dorsett D. Roles of the sister chromatid cohesion apparatus in geneexpression, development, and human syndromes. Chromosoma. 2007 Feb;116(1):1-13.doi: 10.1007/s00412-006-0072-6. Epub 2006 Jul 4. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/16819604) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1783675/)
- Gordillo M, Vega H, Trainer AH, Hou F, Sakai N, Luque R, Kayserili H, BasaranS, Skovby F, Hennekam RC, Uzielli ML, Schnur RE, Manouvrier S, Chang S, Blair E, Hurst JA, Forzano F, Meins M, Simola KO, Raas-Rothschild A, Schultz RA, McDanielLD, Ozono K, Inui K, Zou H, Jabs EW. The molecular mechanism underlying Robertssyndrome involves loss of ESCO2 acetyltransferase activity. Hum Mol Genet. 2008Jul 15;17(14):2172-80. doi: 10.1093/hmg/ddn116. Epub 2008 Apr 14. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/18411254)
- Hou F, Zou H. Two human orthologues of Eco1/Ctf7 acetyltransferases are bothrequired for proper sister-chromatid cohesion. Mol Biol Cell. 2005Aug;16(8):

3908-18. doi: 10.1091/mbc.e04-12-1063. Epub 2005 Jun 15. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/15958495) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1182326/)

- McNairn AJ, Gerton JL. Cohesinopathies: One ring, many obligations. Mutat Res. 2008 Dec 1;647(1-2):103-11. doi: 10.1016/j.mrfmmm.2008.08.010. Epub 2008 Aug 22. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/18786550)
- Resta N, Susca FC, Di Giacomo MC, Stella A, Bukvic N, Bagnulo R, Simone C, Guanti G. A homozygous frameshift mutation in the ESCO2 gene: evidence ofintertissue and interindividual variation in Nmd efficiency. J Cell Physiol. 2006Oct; 209(1):67-73. doi: 10.1002/jcp.20708. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/16775838)
- Schule B, Oviedo A, Johnston K, Pai S, Francke U. Inactivating mutations inESCO2 cause SC phocomelia and Roberts syndrome: no phenotype-genotypecorrelation. Am J Hum Genet. 2005 Dec;77(6):1117-28. doi: 10.1086/498695. Epub2005 Oct 31. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/16380922) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1285169/)
- Vega H, Gordillo M, Jabs EW. ESCO2 Spectrum Disorder. 2006 Apr 18[updated 2020 Mar 26]. In: Adam MP, Feldman J, Mirzaa GM, Pagon RA, Wallace SE,Bean LJH, Gripp KW, Amemiya A, editors. GeneReviews(R) [Internet].Seattle (WA): University of Washington, Seattle; 1993-2024. Available fromhttp://www.ncbi.nlm.nih.gov/books/NBK1153/ Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/20301332)
- Vega H, Trainer AH, Gordillo M, Crosier M, Kayserili H, Skovby F, Uzielli ML,Schnur RE, Manouvrier S, Blair E, Hurst JA, Forzano F, Meins M, Simola KO,Raas-Rothschild A, Hennekam RC, Jabs EW. Phenotypic variability in 49 cases ofESCO2 mutations, including novel missense and codon deletion in theacetyltransferase domain, correlates with ESCO2 expression and establishes theclinical criteria for Roberts syndrome. J Med Genet. 2010 Jan;47(1):30-7. doi:10.1136/jmg.2009.068395. Epub 2009 Jul 1. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/19574259)
- Vega H, Waisfisz Q, Gordillo M, Sakai N, Yanagihara I, Yamada M, van GosligaD, Kayserili H, Xu C, Ozono K, Jabs EW, Inui K, Joenje H. Roberts syndrome iscaused by mutations in ESCO2, a human homolog of yeast ECO1 that is essential forthe establishment of sister chromatid cohesion. Nat Genet. 2005 May;37(5):468-70.doi: 10.1038/ng1548. Epub 2005 Apr 10. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/15821733)
- Whelan G, Kreidl E, Wutz G, Egner A, Peters JM, Eichele G. Cohesinacetyltransferase Esco2 is a cell viability factor and is required for cohesionin pericentric heterochromatin. EMBO J. 2012 Jan 4;31(1):71-82. doi:10.1038/emboj.2011.381. Epub 2011 Nov 18. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/22101327) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3252581/)

Genomic Location

The *ESCO2* gene is found on chromosome 8 (https://medlineplus.gov/genetics/chromosome/8/).

Last updated March 1, 2019