UV-sensitive syndrome

Description

UV-sensitive syndrome is a condition that is characterized by sensitivity to the ultraviolet (UV) rays in sunlight. Even a small amount of sun exposure can cause a sunburn in affected individuals. In addition, these individuals can have freckles, dryness, or changes in coloring (pigmentation) on sun-exposed areas of skin after repeated exposure. Some people with UV-sensitive syndrome have small clusters of enlarged blood vessels just under the skin (telangiectasia), usually on the cheeks and nose. Although UV exposure can cause skin cancers, people with UV-sensitive syndrome do not have an increased risk of developing these forms of cancer compared with the general population.

Frequency

UV-sensitive syndrome appears to be a rare condition; only a small number of affected individuals have been reported in the scientific literature. However, this condition may be underdiagnosed.

Causes

UV-sensitive syndrome can result from mutations in the *ERCC6* gene (also known as the *CSB* gene), the *ERCC8* gene (also known as the *CSA* gene), or the *UVSSA* gene. These genes provide instructions for making proteins that are involved in repairing damaged DNA. DNA can be damaged by UV rays from the sun and by toxic chemicals, radiation, and unstable molecules called free radicals. Cells are usually able to fix DNA damage before it causes problems. If left uncorrected, DNA damage accumulates, which causes cells to malfunction and can lead to cell death.

Cells have several mechanisms to correct DNA damage. The CSB, CSA, and UVSSA proteins are involved in one mechanism that repairs damaged DNA within active genes (those genes undergoing gene transcription, the first step in protein production). When DNA in active genes is damaged, the enzyme that carries out gene transcription (RNA polymerase) gets stuck, and the process stalls. Researchers think that the CSB, CSA, and UVSSA proteins help remove RNA polymerase from the damaged site, so the DNA can be repaired.

Mutations in the *ERCC6*, *ERCC8*, or *UVSSA* genes lead to the production of an

abnormal protein or the loss of the protein. If any of these proteins is not functioning normally, skin cells cannot repair DNA damage caused by UV rays, and transcription of damaged genes is blocked. However, it is unclear exactly how abnormalities in these proteins cause the signs and symptoms of UV-sensitive syndrome.

Learn more about the genes associated with UV-sensitive syndrome

- ERCC6
- ERCC8
- UVSSA

Inheritance

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- Ultraviolet sensitive syndrome
- UVSS

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: UV-sensitive syndrome 1 (https://www.ncbi.nlm.nih.gov/gtr/conditions/C3551173/)
- Genetic Testing Registry: UV-sensitive syndrome 2 (https://www.ncbi.nlm.nih.gov/gtr/conditions/C3553298/)
- Genetic Testing Registry: UV-sensitive syndrome 3 (https://www.ncbi.nlm.nih.gov/gtr/conditions/C3553328/)

Genetic and Rare Diseases Information Center

- UV-sensitive syndrome (https://rarediseases.info.nih.gov/diseases/10947/index)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (https://rarediseases.org/)

Catalog of Genes and Diseases from OMIM

- UV-SENSITIVE SYNDROME 1; UVSS1 (https://omim.org/entry/600630)
- UV-SENSITIVE SYNDROME 2; UVSS2 (https://omim.org/entry/614621)
- UV-SENSITIVE SYNDROME 3; UVSS3 (https://omim.org/entry/614640)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28uv-sensitive+syndrome%5BTIAB%5D%29+OR+%28UVSS%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

References

- Horibata K, Iwamoto Y, Kuraoka I, Jaspers NG, Kurimasa A, Oshimura M,Ichihashi M, Tanaka K. Complete absence of Cockayne syndrome group B gene productgives rise to UV-sensitive syndrome but not Cockayne syndrome. Proc Natl Acad SciU S A. 2004 Oct 26;101(43):15410-5. doi: 10.1073/pnas.0404587101. Epub 2004 Oct14. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/15486090) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC524447/)
- Itoh T, Fujiwara Y, Ono T, Yamaizumi M. UVs syndrome, a new general categoryof photosensitive disorder with defective DNA repair, is distinct from xerodermapigmentosum variant and rodent complementation group I. Am J Hum Genet. 1995Jun;56(6):1267-76. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/7539208) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1801097/)
- Itoh T, Yamaizumi M, Ichihashi M, Hiro-Oka M, Matsui T, Matsuno M, Ono T. Clinical characteristics of three patients with UVs syndrome, a photosensitivedisorder with defective DNA repair. Br J Dermatol. 1996 Jun;134(6): 1147-50. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/8763445)
- Nardo T, Oneda R, Spivak G, Vaz B, Mortier L, Thomas P, Orioli D, Laugel V,Stary A, Hanawalt PC, Sarasin A, Stefanini M. A UV-sensitive syndrome patientwith a specific CSA mutation reveals separable roles for CSA in response to UVand oxidative DNA damage. Proc Natl Acad Sci U S A. 2009 Apr 14;106(15):6209-14.doi: 10.1073/pnas.0902113106. Epub 2009 Mar 27. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/19329487) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2667150/)
- Spivak G, Itoh T, Matsunaga T, Nikaido O, Hanawalt P, Yamaizumi M.Ultraviolet-sensitive syndrome cells are defective in transcription-coupledrepair of cyclobutane pyrimidine dimers. DNA Repair (Amst). 2002 Aug6;1(8):629-43. doi: 10.1016/s1568-7864(02)00056-3. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/12509286)
- Zhang X, Horibata K, Saijo M, Ishigami C, Ukai A, Kanno S, Tahara H, NeilanEG, Honma M, Nohmi T, Yasui A, Tanaka K. Mutations in UVSSA cause UV-sensitivesyndrome and destabilize ERCC6 in transcription-coupled DNA repair. Nat

Genet.2012 May;44(5):593-7. doi: 10.1038/ng.2228. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/22466612)

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