PACS1 Related Syndrome

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PACS1 gene

phosphofurin acidic cluster sorting protein 1

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Normal Function

The PACS1 gene provides instructions for making a protein called phosphofurin acidic cluster sorting protein 1 (PACS1). The PACS1 protein is found in a complex network of membranes known as the trans-Golgi network, which sorts proteins and other molecules and sends them to their intended destinations inside or outside the cell. Within the trans-Golgi network, this protein helps transport certain molecules and proteins. The PACS1 protein is most active during development before birth.

Health Conditions Related to Genetic Changes

PACS1 syndrome

At least two mutations in the PACS1 gene have been found to cause PACS1 syndrome. This condition is characterized by intellectual disability, speech and language problems, and a distinct facial appearance. Many affected individuals have additional neurological, behavioral, and health problems. The most common mutation, which occurs in nearly everyone with PACS1 syndrome, results in the production of a protein with the protein building block (amino acid) arginine replaced with the amino acid tryptophan at position 203 (written as Arg203Trp or R203W).PACS1 gene mutations are thought to impair the protein's ability to aid in the transport of molecules and proteins. Such an impairment likely results in the accumulation or misplacement of these substances within cells. The accumulated molecules and proteins may interfere with the function of the protein produced from the normal copy of the PACS1 gene, further disrupting the placement of these substances.Research suggests that impaired PACS1 protein function disrupts normal development of structures in the face, leading to a distinct facial appearance. It is likely that the development of other body systems are similarly affected by impaired PACS1 protein function, leading to other signs and symptoms of PACS1 syndrome, but more research is needed to understand the mechanisms.

More About This Health Condition

Other Names for This Gene

cytosolic sorting protein PACS-1, human FLJ10209 KIAA1175 PACS-1 phosphofurin acidic cluster sorting protein 1, human

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

Tests of PACS1

Scientific Articles on PubMed

PubMed

Catalog of Genes and Diseases from OMIM

PHOSPHOFURIN ACIDIC CLUSTER SORTING PROTEIN 1; PACS1

Gene and Variant Databases

NCBI Gene

ClinVar

References

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Citation on PubMed

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Genomic LocationThe PACS1 gene is found on chromosome 11.

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