# PTPN22 gene

protein tyrosine phosphatase non-receptor type 22

## Normal Function

The *PTPN22* gene provides instructions for making a protein that belongs to the PTP ( protein tyrosine phosphatases) family. PTP proteins play a role in regulating a process called signal transduction. In signal transduction, the protein relays signals from outside the cell to the cell nucleus. These signals instruct the cell to grow and divide or to mature and take on specialized functions.

The PTPN22 protein is involved in signaling that helps control the activity of immune system cells called T cells. T cells identify foreign substances and defend the body against infection.

## Health Conditions Related to Genetic Changes

Vitiligo

Studies have associated the R620W variation in the *PTPN22* gene with an increased risk of vitiligo, an autoimmune condition that results in patchy changes in skin coloring ( pigmentation).

As with other autoimmune disorders, this variation likely affects the activity of the PTPN22 protein, making it more difficult for the body to control the immune system and prevent it from attacking its own tissues. While the pigment loss associated with vitiligo results from the immune system attacking pigment-producing cells (melanocytes) in the skin, it is unclear what specific circumstances trigger the immune system to do so. The condition probably results from a combination of genetic and environmental factors, most of which have not been identified.

Alopecia areata

MedlinePlus Genetics provides information about Alopecia areata

Autoimmune Addison disease

MedlinePlus Genetics provides information about Autoimmune Addison disease

Graves' disease

MedlinePlus Genetics provides information about Graves' disease

Hashimoto's disease

MedlinePlus Genetics provides information about Hashimoto's disease

Idiopathic inflammatory myopathy

MedlinePlus Genetics provides information about Idiopathic inflammatory myopathy

Juvenile idiopathic arthritis

MedlinePlus Genetics provides information about Juvenile idiopathic arthritis

Rheumatoid arthritis

MedlinePlus Genetics provides information about Rheumatoid arthritis

Systemic lupus erythematosus

MedlinePlus Genetics provides information about Systemic lupus erythematosus

Systemic scleroderma

MedlinePlus Genetics provides information about Systemic scleroderma

Type 1 diabetes

MedlinePlus Genetics provides information about Type 1 diabetes

Autoimmune disorders

Studies have associated a variation in the *PTPN22* gene with an increased risk of several autoimmune disorders. Autoimmune disorders occur when the immune system malfunctions and attacks the body's tissues and organs. These disorders include type 1 diabetes, rheumatoid arthritis, Hashimoto thyroiditis, Graves disease, and systemic lupus erythematosus.

The *PTPN22* gene variation associated with autoimmune disorders changes the protein building block (amino acid) arginine to the amino acid tryptophan at position 620 in the PTPN22 protein sequence, written as Arg620Trp or R620W. This variation likely affects the activity of the PTPN22 protein, making it more difficult for the body to control inflammation and prevent the immune system from attacking its own tissues.

## Other Names for This Gene

- hematopoietic cell protein-tyrosine phosphatase 70Z-PEP
- lymphoid phosphatase
- lymphoid-specific protein tyrosine phosphatase

- LYP
- LYP1
- LYP2
- PEP
- PEST-domain phosphatase
- protein tyrosine phosphatase, non-receptor type 22 (lymphoid)
- protein tyrosine phosphatase, non-receptor type 8
- PTN22_HUMAN
- PTPN8
- tyrosine-protein phosphatase non-receptor type 22

## Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of PTPN22 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=26191[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28PTPN22%5BTI%5D%29+OR+%28protein+tyrosine+phosphatase,+non-receptor+type+22%5BTI%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- PROTEIN TYROSINE PHOSPHATASE, NONRECEPTOR-TYPE, 22; PTPN22 (https://omim.org/entry/600716)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/26191)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=PTPN22[gene])

## References


- Arechiga AF, Habib T, He Y, Zhang X, Zhang ZY, Funk A, Buckner JH. Cuttingedge: the PTPN22 allelic variant associated with autoimmunity impairs B cellsignaling. J Immunol. 2009 Mar 15;182(6):3343-7. doi: 10.4049/jimmunol.0713370. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/19265110) or Free article on PubMed

Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2797545/)

- LaBerge GS, Bennett DC, Fain PR, Spritz RA. PTPN22 is genetically associatedwith risk of generalized vitiligo, but CTLA4 is not. J Invest Dermatol. 2008Jul;128(7):1757-62. doi: 10.1038/sj.jid.5701233. Epub 2008 Jan 17. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/18200060)
- Laberge GS, Birlea SA, Fain PR, Spritz RA. The PTPN22-1858C>T (R620W) functional polymorphism is associated with generalized vitiligo in the Romanianpopulation. Pigment Cell Melanoma Res. 2008 Apr;21(2):206-8. doi:10.1111/j.1755-148X.2008.00443.x. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/18426414)
- Lee YH, Rho YH, Choi SJ, Ji JD, Song GG, Nath SK, Harley JB. The PTPN22 C1858Tfunctional polymorphism and autoimmune diseases--a meta-analysis. Rheumatology(Oxford). 2007 Jan;46(1):49-56. doi: 10.1093/rheumatology/kel170. Epub 2006 Jun7. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/16760194)
- Pradhan V, Borse V, Ghosh K. PTPN22 gene polymorphisms in autoimmune diseaseswith special reference to systemic lupus erythematosus disease susceptibility. JPostgrad Med. 2010 Jul-Sep;56(3):239-42. doi: 10.4103/0022-3859.68651. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/20739780)
- Spritz RA. The genetics of generalized vitiligo and associated autoimmunediseases. Pigment Cell Res. 2007 Aug;20(4):271-8. doi:10.1111/j.1600-0749.2007.00384.x. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/17630960)
- Vang T, Miletic AV, Bottini N, Mustelin T. Protein tyrosine phosphatase PTPN22in human autoimmunity. Autoimmunity. 2007 Sep;40(6):453-61. doi:10.1080/08916930701464897. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/17729039)

## Genomic Location

The *PTPN22* gene is found on chromosome 1 (https://medlineplus.gov/genetics/chromosome/1/).

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