



INS gene

insulin

Normal Function

The *INS* gene provides instructions for producing the hormone insulin, which is necessary for the control of glucose levels in the blood. Glucose is a simple sugar and the primary energy source for most cells in the body.

Insulin is produced in a precursor form called proinsulin, which consists of a single chain of protein building blocks (amino acids). The proinsulin chain is cut (cleaved) to form individual pieces called the A and B chains, which are joined together by connections called disulfide bonds to form insulin.

Health Conditions Related to Genetic Changes

Permanent neonatal diabetes mellitus

At least 10 mutations in the *INS* gene have been identified in people with permanent neonatal diabetes mellitus. Individuals with this condition often have a low birth weight and develop increased blood sugar (hyperglycemia) within the first 6 months of life.

INS gene mutations that cause permanent neonatal diabetes mellitus change single protein building blocks (amino acids) in the protein sequence. These mutations are believed to disrupt the cleavage of the proinsulin chain or the binding of the A and B chains to form insulin, leading to impaired blood sugar control.

Type 1 diabetes

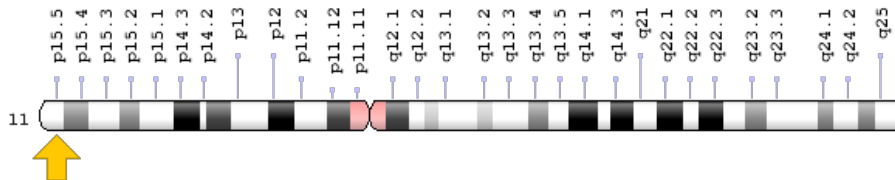
Other disorders

Mutations in the *INS* gene can also cause other disorders involving insulin production and blood sugar control. Some individuals with *INS* gene mutations have increased levels of proinsulin in their blood (hyperproinsulinemia) and may also have impaired blood sugar control. *INS* gene mutations are also associated with a disorder called maturity-onset diabetes of the young (MODY). This term refers to hereditary forms of relatively mild diabetes mellitus caused by changes in single genes.

Chromosomal Location

Cytogenetic Location: 11p15.5, which is the short (p) arm of chromosome 11 at position 15.5

Molecular Location: base pairs 2,159,779 to 2,161,209 on chromosome 11 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- IDDM2
- ILPR
- INS_HUMAN
- insulin preproprotein
- IRDN
- MODY10
- proinsulin

Additional Information & Resources

Educational Resources

- The Genetic Landscape of Diabetes (NCBI, 2004): Insulin Synthesis
<https://www.ncbi.nlm.nih.gov/books/NBK1671/#A622>

Clinical Information from GeneReviews

- Permanent Neonatal Diabetes Mellitus
<https://www.ncbi.nlm.nih.gov/books/NBK1447>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28INS%5BTI%5D%29+OR+%28insulin%5BTI%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+review%5Bpt%5D+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D>

Catalog of Genes and Diseases from OMIM

- INSULIN
<http://omim.org/entry/176730>
- MATURITY-ONSET DIABETES OF THE YOUNG, TYPE 10
<http://omim.org/entry/613370>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_INS.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=INS%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report#!/hgnc_id/HGNC:6081
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:3630>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/3630>
- UniProt
<https://www.uniprot.org/uniprot/P01308>

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