

## GEPHE SUMMARY

<p>HBS1L-MYB (<a href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=~HBS1L-MYB^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=~HBS1L-MYB^#gephebase-summary-title</a>)</p> <p>Published</p>	<p>Gephebase Gene</p> <p>Entry Status</p>	<p>GP00001608</p> <p>Prigent</p>	<p>GepheID</p> <p>Main curator</p>
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## PHENOTYPIC CHANGE

<p>Physiology (<a href="https://www.gephebase.org/search-criteria?/and+Trait+Category=~Physiology^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait+Category=~Physiology^#gephebase-summary-title</a>)</p> <p>Hematopoiesis (mean blood corpuscular hemoglobin) (<a href="https://www.gephebase.org/search-criteria?/and+Trait=~Hematopoiesis+(mean+blood+corpuscular+hemoglobin)^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait=~Hematopoiesis+(mean+blood+corpuscular+hemoglobin)^#gephebase-summary-title</a>)</p> <p>Human - Estonia Biobank</p> <p>Human - Estonia Biobank</p> <p>Unknown</p> <p>Intraspecific (<a href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=~Intraspecific^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=~Intraspecific^#gephebase-summary-title</a>)</p>	<p>Trait Category</p> <p>Trait</p> <p>Trait State in Taxon A</p> <p>Trait State in Taxon B</p> <p>Ancestral State</p> <p>Taxonomic Status</p>	<p>Taxon A</p> <p>Latin Name</p> <p>Homo sapiens (<a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Homo+sapiens^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Homo+sapiens^#gephebase-summary-title</a>)</p> <p>Common Name</p> <p>human</p> <p>Synonyms</p> <p>human; man; Homo sapiens Linnaeus, 1758; Home sapiens; Homo sampiens; Homo sapeins; Homo sapian; Homo sapians; Homo sapien; Homo sapience; Homo sapiense; Homo sapients; Homo sapines; Homo spaiens; Homo spiens; Humo sapiens</p> <p>Rank</p> <p>species</p> <p>Lineage</p> <p>cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Euarchontoglires; Primates; Haplorrhini; Simiiformes; Catarrhini; Hominoidea; Hominidae; Homininae; Homo</p> <p>Parent</p> <p>Homo () - (Rank: genus) (<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605</a>)</p> <p>NCBI Taxonomy ID</p> <p>9606 (<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606</a>)</p> <p>is Taxon A an Intraspecies?</p> <p>Yes</p> <p>Taxon A Description</p> <p>Human - Estonia Biobank</p>	<p>Taxon B</p> <p>Latin Name</p> <p>Homo sapiens (<a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Homo+sapiens^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Homo+sapiens^#gephebase-summary-title</a>)</p> <p>Common Name</p> <p>human</p> <p>Synonyms</p> <p>human; man; Homo sapiens Linnaeus, 1758; Home sapiens; Homo sampiens; Homo sapeins; Homo sapian; Homo sapians; Homo sapien; Homo sapience; Homo sapiense; Homo sapients; Homo sapines; Homo spaiens; Homo spiens; Humo sapiens</p> <p>Rank</p> <p>species</p> <p>Lineage</p> <p>cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Euarchontoglires; Primates; Haplorrhini; Simiiformes; Catarrhini; Hominoidea; Hominidae; Homininae; Homo</p> <p>Parent</p> <p>Homo () - (Rank: genus) (<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605</a>)</p> <p>NCBI Taxonomy ID</p> <p>9606 (<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606</a>)</p> <p>is Taxon B an Intraspecies?</p> <p>Yes</p> <p>Taxon B Description</p> <p>Human - Estonia Biobank</p>
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## GENOTYPIC CHANGE

<p>HBS1L</p> <p>ERFS; HBS1; EF-1a; eRF3c; HSPC276; KIAA1038</p> <p>9606.ENSPO0000356811 (<a href="http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPO0000356811">http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPO0000356811</a>)</p> <p>Sequence Similarities</p> <p>Belongs to the TRAFAC class translation factor GTPase superfamily. Classic translation factor GTPase family.</p> <p>GO - Molecular Function</p>	<p>Generic Gene Name</p> <p>Synonyms</p> <p>String</p>	<p>Q9Y450 (<a href="http://www.uniprot.org/uniprot/Q9Y450">http://www.uniprot.org/uniprot/Q9Y450</a>)</p> <p>()</p>	<p>UniProtKB Homo sapiens</p> <p>GenebankID or UniProtKB</p>
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GO:0005525 : GTP binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0005525>)  
GO:0003924 : GTPase activity (<https://www.ebi.ac.uk/QuickGO/term/GO:0003924>)  
GO:0003746 : translation elongation factor activity  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0003746>)

GO - Biological Process

GO:0007165 : signal transduction (<https://www.ebi.ac.uk/QuickGO/term/GO:0007165>)  
GO:0006412 : translation (<https://www.ebi.ac.uk/QuickGO/term/GO:0006412>)  
GO:0043928 : exonucleolytic nuclear-transcribed mRNA catabolic process involved in  
deadenylation-dependent decay (<https://www.ebi.ac.uk/QuickGO/term/GO:0043928>)

GO - Cellular Component

GO:0005829 : cytosol (<https://www.ebi.ac.uk/QuickGO/term/GO:0005829>)  
GO:0016020 : membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0016020>)  
GO:0070062 : extracellular exosome (<https://www.ebi.ac.uk/QuickGO/term/GO:0070062>)

Presumptive Null

No (<https://www.gephebase.org/search-criteria?/and+Presumptive Null=^No^#gephebase-summary-title>)

Molecular Type

Cis-regulatory (<https://www.gephebase.org/search-criteria?/and+Molecular Type=^Cis-regulatory^#gephebase-summary-title>)

Aberration Type

SNP (<https://www.gephebase.org/search-criteria?/and+Aberration Type=^SNP^#gephebase-summary-title>)

Molecular Details of the Mutation

A>G at the associated SNP. Variants in MEP/erythroid-specific elements are putative functional variants

Experimental Evidence

Association Mapping (<https://www.gephebase.org/search-criteria?/and+Experimental Evidence=^Association Mapping^#gephebase-summary-title>)

Main Reference

Comprehensive population-based genome sequencing provides insight into hematopoietic regulatory mechanisms. (2017) (<https://pubmed.ncbi.nlm.nih.gov/28031487>)

Authors

Guo MH; Nandakumar SK; Ulirsch JC; Zekavat SM; Buenrostro JD; Natarajan P; Salem RM; Chiarle R; Mitt M; Kals M; PÄärn K; Fischer K; Milani L; MÄägi R; Palta P; Gabriel SB; Metspalu A; Lander ES; Kathiresan S; Hirschhorn JN; Esko T; Sankaran VG

Abstract

Genetic variants affecting hematopoiesis can influence commonly measured blood cell traits. To identify factors that affect hematopoiesis, we performed association studies for blood cell traits in the population-based Estonian Biobank using high-coverage whole-genome sequencing (WGS) in 2,284 samples and SNP genotyping in an additional 14,904 samples. Using up to 7,134 samples with available phenotype data, our analyses identified 17 associations across 14 blood cell traits. Integration of WGS-based fine-mapping and complementary epigenomic datasets provided evidence for causal mechanisms at several loci, including at a previously undiscovered basophil count-associated locus near the master hematopoietic transcription factor CEBPA. The fine-mapped variant at this basophil count association near CEBPA overlapped an enhancer active in common myeloid progenitors and influenced its activity. In situ perturbation of this enhancer by CRISPR/Cas9 mutagenesis in hematopoietic stem and progenitor cells demonstrated that it is necessary for and specifically regulates CEBPA expression during basophil differentiation. We additionally identified basophil count-associated variation at another more pleiotropic myeloid enhancer near GATA2, highlighting regulatory mechanisms for ordered expression of master hematopoietic regulators during lineage specification. Our study illustrates how population-based genetic studies can provide key insights into poorly understood cell differentiation processes of considerable physiologic relevance.

Additional References

## RELATED GEPHE

Related Genes

12 (ARHGEF3, BAK1, CCAAT-enhancer-binding protein alpha (CEBPA), F2RL2, GATA-binding protein 2 (GATA2), JAK2, JMJD1C, LPAR1, PIK3CG, PSMD13, TMPRSS6, WDR66) (<https://www.gephebase.org/search-criteria?/or+Taxon ID=^9606^/and+Trait=Hematopoiesis/and+groupHaplotypes=true#gephebase-summary-title>)

Related Haplotypes

3 (<https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^HBS1L-MYB^/and+Taxon ID=^9606^/or+Gene Gephebase=^HBS1L-MYB^/and+Taxon ID=^9606^#gephebase-summary-title>)

## EXTERNAL LINKS

## COMMENTS