

GEPHE SUMMARY

|                                                                                                                                                                                                                      |                |            |              |
|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|----------------|------------|--------------|
| ABCC11 ( <a href="https://www.gephebase.org/search-criteria?/and+GeneGephebase=~ABCC11^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+GeneGephebase=~ABCC11^#gephebase-summary-title</a> ) | Gephebase Gene | GP00000010 | GepheID      |
|                                                                                                                                                                                                                      | Entry Status   | Martin     | Main curator |
|                                                                                                                                                                                                                      | Published      |            |              |

PHENOTYPIC CHANGE

|                                                                                                                                                                                                                                                                                                                                                                                  |  |                                                                                                                                                                                                                                                                                                                                                                                  |  |
|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|--|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|--|
| Physiology ( <a href="https://www.gephebase.org/search-criteria?/and+TraitCategory=~Physiology^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+TraitCategory=~Physiology^#gephebase-summary-title</a> )                                                                                                                                                 |  | Trait Category                                                                                                                                                                                                                                                                                                                                                                   |  |
| Apocrine secretion (ear wax type ; axillary odor) ( <a href="https://www.gephebase.org/search-criteria?/and+Trait=~Apocrine secretion (ear wax type ; axillary odor)^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait=~Apocrine secretion (ear wax type ; axillary odor)^#gephebase-summary-title</a> )                                            |  | Trait                                                                                                                                                                                                                                                                                                                                                                            |  |
| Homo sapiens - wet earwax                                                                                                                                                                                                                                                                                                                                                        |  | Trait State in Taxon A                                                                                                                                                                                                                                                                                                                                                           |  |
| Homo sapiens - dry earwax                                                                                                                                                                                                                                                                                                                                                        |  | Trait State in Taxon B                                                                                                                                                                                                                                                                                                                                                           |  |
| Data not curated                                                                                                                                                                                                                                                                                                                                                                 |  | Ancestral State                                                                                                                                                                                                                                                                                                                                                                  |  |
| Intraspecific ( <a href="https://www.gephebase.org/search-criteria?/and+TaxonomicStatus=~Intraspecific^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+TaxonomicStatus=~Intraspecific^#gephebase-summary-title</a> )                                                                                                                                    |  | Taxonomic Status                                                                                                                                                                                                                                                                                                                                                                 |  |
| Taxon A                                                                                                                                                                                                                                                                                                                                                                          |  | Taxon B                                                                                                                                                                                                                                                                                                                                                                          |  |
| Homo sapiens<br>( <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> )                                                                                                                              |  | Homo sapiens<br>( <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> )                                                                                                                              |  |
| human                                                                                                                                                                                                                                                                                                                                                                            |  | human                                                                                                                                                                                                                                                                                                                                                                            |  |
| 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                                                                                                                                                   |  | 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                                                                                                                                                   |  |
| species                                                                                                                                                                                                                                                                                                                                                                          |  | species                                                                                                                                                                                                                                                                                                                                                                          |  |
| 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 |  | 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 |  |
| Homo () - (Rank: genus)<br>( <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> )                                                                                                                                                                                                 |  | Homo () - (Rank: genus)<br>( <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> )                                                                                                                                                                                                 |  |
| 9606<br>( <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> )                                                                                                                                                                                                                    |  | 9606<br>( <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> )                                                                                                                                                                                                                    |  |
| No                                                                                                                                                                                                                                                                                                                                                                               |  | No                                                                                                                                                                                                                                                                                                                                                                               |  |

GENOTYPIC CHANGE

|                                                                                                                                                                                                                                 |                   |                                                                                                                        |
|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-------------------|------------------------------------------------------------------------------------------------------------------------|
| ABCC11                                                                                                                                                                                                                          | Generic Gene Name | UniProtKB Homo sapiens                                                                                                 |
|                                                                                                                                                                                                                                 | Synonyms          | Q96J66 ( <a href="http://www.uniprot.org/uniprot/Q96J66">http://www.uniprot.org/uniprot/Q96J66</a> )                   |
|                                                                                                                                                                                                                                 | String            | AF367202 ( <a href="https://www.ncbi.nlm.nih.gov/nuccore/AF367202">https://www.ncbi.nlm.nih.gov/nuccore/AF367202</a> ) |
| 9606.ENSPO0000349017<br>( <a href="http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPO0000349017">http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPO0000349017</a> ) |                   |                                                                                                                        |
| Belongs to the ABC transporter superfamily. ABCC family. Conjugate transporter (TC 3.A.1.208) subfamily.                                                                                                                        |                   |                                                                                                                        |
| GO:0005524 : ATP binding ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0005524">https://www.ebi.ac.uk/QuickGO/term/GO:0005524</a> )                                                                                          |                   |                                                                                                                        |
| GO:0042626 : ATPase activity, coupled to transmembrane movement of substances                                                                                                                                                   |                   |                                                                                                                        |

(<https://www.ebi.ac.uk/QuickGO/term/GO:0042626>)  
GO:0043225 : ATPase-coupled anion transmembrane transporter activity  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043225>)  
GO:0008514 : organic anion transmembrane transporter activity  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0008514>)  
GO:0015216 : purine nucleotide transmembrane transporter activity  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0015216>)

GO - Biological Process

GO:0015711 : organic anion transport (<https://www.ebi.ac.uk/QuickGO/term/GO:0015711>)  
GO:0015865 : purine nucleotide transport  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0015865>)  
GO:0055085 : transmembrane transport  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0055085>)

GO - Cellular Component

GO:0005886 : plasma membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0005886>)  
GO:0016020 : membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0016020>)  
GO:0030659 : cytoplasmic vesicle membrane  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030659>)  
GO:0070062 : extracellular exosome (<https://www.ebi.ac.uk/QuickGO/term/GO:0070062>)  
GO:0005887 : integral component of plasma membrane  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0005887>)  
GO:0005774 : vacuolar membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0005774>)

Presumptive Null

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

Molecular Type

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

Aberration Type

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

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

1 a.a. polymorphism considered best candidate - c.538G>A - G180R in the ATP-binding cassette

Experimental Evidence

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

|            | Taxon A | Taxon B | Position |
|------------|---------|---------|----------|
| Codon      | -       | -       | -        |
| Amino-acid | -       | -       | -        |

Main Reference

A SNP in the ABCC11 gene is the determinant of human earwax type. (2006) (<https://pubmed.ncbi.nlm.nih.gov/16444273>)

Authors

Yoshiura K; Kinoshita A; Ishida T; Ninokata A; Ishikawa T; Kaname T; Bannai M; Tokunaga K; Sonoda S; Komaki R; Ihara M; Saenko VA; Alipov GK; Sekine I; Komatsu K; Takahashi H; Nakashima M; Sosonkina N; Mapendano CK; Ghadami M; Nomura M; Liang DS; Miwa N; Kim DK; Garidkhuu A; Natsume N; Ohta T; Tomita H; Kaneko A; Kikuchi M; Russomando G; Hirayama K; Ishibashi M; Takahashi A; Saitou N; Murray JC; Saito S; Nakamura Y; Niikawa N

Abstract

Human earwax consists of wet and dry types. Dry earwax is frequent in East Asians, whereas wet earwax is common in other populations. Here we show that a SNP, 538G --> A (rs17822931), in the ABCC11 gene is responsible for determination of earwax type. The AA genotype corresponds to dry earwax, and GA and GG to wet type. A 27-bp deletion in ABCC11 exon 29 was also found in a few individuals of Asian ancestry. A functional assay demonstrated that cells with allele A show a lower excretory activity for cGMP than those with allele G. The allele A frequency shows a north-south and east-west downward geographical gradient; worldwide, it is highest in Chinese and Koreans, and a common dry-type haplotype is retained among various ethnic populations. These suggest that the allele A arose in northeast Asia and thereafter spread through the world. The 538G --> A SNP is the first example of DNA polymorphism determining a visible genetic trait.

Additional References

A functional ABCC11 allele is essential in the biochemical formation of human axillary odor. (2010) (<https://pubmed.ncbi.nlm.nih.gov/19710689>)

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

