Resource Summary Report

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Human Intermediate Filament Database

RRID:SCR_007744
Type: Tool

Proper Citation

Human Intermediate Filament Database (RRID:SCR_007744)

Resource Information

URL: http://www.interfil.org

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Description: The Human Intermediate Filament Database is a continuously updated review of the intermediate filament field. It is hoped that users will contribute to the development and expansion of the database on a regular basis. Contributions may include novel variants, new patients with previously discovered sequence and allelic variants. Suggestions on ways to improve the database are also welcome. The entire database can be searched through the Browse and Search options. A number of different parameters can be used to search the database including unique identifier, intermediate filament, disease DNA variations, amino acid variations, domain, date accepted, author and abstract. Output from the search is returned in a table containing all the pertinent cross referenced information. Multiple sequence alignment can also be performed via the CLUSTALW program to determine cDNA or protein sequence conservation. The database is linked to multiple other resources including NCBI RefSeq, PDB, OMIM, UCSC genome browser, NCBI Gene, HomoloGene, PubMed and HGNC. In the case of HGNC, reciprocal links are also available from HGNC that links to Human Intermediate Filament Database. Due to the protein centric nature of the Human Intermediate Filament Database and the gene centric nature of HGNC, a HGNC record will potentially link to multiple records in this database due to the presence of alternative splicing. In such an event, the Human Intermediate Filament Database will present to the user a list of all the protein records resulting from the HGNC gene record. The database uses Jalview and Jmol applets for the visualization of multiple sequence alignment and structure respectively. The database contains information on disease phenotypes of a variety of different intermediate filament related diseases.

Synonyms: InterFil

Resource Type: data or information resource, database

Keywords: FASEB list

Resource Name: Human Intermediate Filament Database

Resource ID: SCR_007744

Alternate IDs: nif-0000-03034, r3d100012359

Alternate URLs: https://doi.org/10.17616/R3T66K

Record Creation Time: 20220129T080243+0000

Record Last Update: 20260912T200151+0000

Ratings and Alerts

No rating or validation information has been found for Human Intermediate Filament Database.

No alerts have been found for Human Intermediate Filament Database.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 62 mentions in open access literature.

Listed below are recent publications. The full list is available at T1D .

Ho M, et al. (2022) Update of the keratin gene family: evolution, tissue-specific expression patterns, and relevance to clinical disorders. Human genomics, 16(1), 1.

Elsnicova B, et al. (2022) Desmin Knock-Out Cardiomyopathy: A Heart on the Verge of Metabolic Crisis. International journal of molecular sciences, 23(19).

Lehmann SM, et al. (2021) Growth, lifetime, directional movement and myosin-dependent motility of mutant keratin granules in cultured cells. Scientific reports, 11(1), 2379.

Paduano F, et al. (2021) A Familial Form of Epidermolysis Bullosa Simplex Associated with a Pathogenic Variant in KRT5. Genes, 12(10).

Sjöqvist M, et al. (2021) From structural resilience to cell specification - Intermediate filaments as regulators of cell fate. FASEB journal : official publication of the Federation of American Societies for Experimental Biology, 35(1), e21182.

Bchetnia M, et al. (2021) Genetic burden linked to founder effects in Saguenay-Lac-Saint-Jean illustrates the importance of genetic screening test availability. Journal of medical genetics, 58(10), 653.

Yu Y, et al. (2021) Epidermolysis Bullosa in Chinese Patients: Genetic Analysis and Mutation Landscape in 57 Pedigrees and Sporadic Cases. Acta dermato-venereologica,

101(7), adv00503.

Sapra KT, et al. (2021) Bend, Push, Stretch: Remarkable Structure and Mechanics of Single Intermediate Filaments and Meshworks. Cells, 10(8).

Has C, et al. (2020) Clinical practice guidelines for laboratory diagnosis of epidermolysis bullosa. The British journal of dermatology, 182(3), 574.

Diociaiuti A, et al. (2020) First Case of KRT2 Epidermolytic Nevus and Novel Clinical and Genetic Findings in 26 Italian Patients with Keratinopathic Ichthyoses. International journal of molecular sciences, 21(20).

Mohammed F, et al. (2020) Molecular mechanism of intermediate filament recognition by plakin proteins. Biochimica et biophysica acta. Molecular cell research, 1867(11), 118801.

Bchetnia M, et al. (2020) Generation of a human induced pluripotent stem cell line (UQACi001-A) from a severe epidermolysis bullosa simplex patient with the heterozygous mutation p.R125S in the KRT14 gene. Stem cell research, 44, 101748.

Eiber N, et al. (2020) Lack of Desmin in Mice Causes Structural and Functional Disorders of Neuromuscular Junctions. Frontiers in molecular neuroscience, 13, 567084.

Eriksson JE, et al. (2020) Harmful vimentin manifests itself as multiorgan failure. European journal of human genetics : EJHG, 28(9), 1139.

Lee CH, et al. (2020) Structure-Function Analyses of a Keratin Heterotypic Complex Identify Specific Keratin Regions Involved in Intermediate Filament Assembly. Structure (London, England : 1993), 28(3), 355.

Klymkowsky MW, et al. (2019) Filaments and phenotypes: cellular roles and orphan effects associated with mutations in cytoplasmic intermediate filament proteins. F1000Research, 8.

Tryon RK, et al. (2019) A homozygous frameshift variant in the KRT5 gene is compatible with life and results in severe recessive epidermolysis bullosa simplex. JAAD case reports, 5(7), 576.

Ruppert T, et al. (2019) AAV-mediated cardiac gene transfer of wild-type desmin in mouse models for recessive desminopathies. Gene therapy, 27(10-11), 516.

Borská R, et al. (2019) Inherited ichthyoses: molecular causes of the disease in Czech patients. Orphanet journal of rare diseases, 14(1), 92.

Battaglia RA, et al. (2019) Site-specific phosphorylation and caspase cleavage of GFAP are new markers of Alexander disease severity. eLife, 8.