

Resource Summary Report

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PEDCHECK

RRID:SCR_009322

Type: Tool

Proper Citation

PEDCHECK (RRID:SCR_009322)

Resource Information

URL: <http://watson.hgen.pitt.edu/register>

Proper Citation: PEDCHECK (RRID:SCR_009322)

Description: THIS RESOURCE IS NO LONGER IN SERVICE. Documented on May 16,20023. Software application for identifying all Mendelian inconsistencies in pedigree data. (entry from Genetic Analysis Software)

Abbreviations: PEDCHECK

Resource Type: software application, software resource

Keywords: gene, genetic, genomic, c, c++, unix, (sunos/solaris/compaq-alpha/sgi-irix/..)

Availability: THIS RESOURCE IS NO LONGER IN SERVICE

Resource Name: PEDCHECK

Resource ID: SCR_009322

Alternate IDs: nlx_154516

Record Creation Time: 20220129T080252+0000

Record Last Update: 20260727T040458+0000

Ratings and Alerts

No rating or validation information has been found for PEDCHECK.

No alerts have been found for PEDCHECK.

Data and Source Information

Usage and Citation Metrics

We found 75 mentions in open access literature.

Listed below are recent publications. The full list is available at [nidm-terms](#) .

Aydin A, et al. (2024) ADAM19 cleaves the PTH receptor and associates with brachydactyly type E. Life science alliance, 7(4).

Bezamat M, et al. (2024) Family-based GWAS for dental class I malocclusion and clefts. BMC oral health, 24(1), 665.

Abdel-Salam GMH, et al. (2023) Biallelic MAD2L1BP (p31comet) mutation is associated with mosaic aneuploidy and juvenile granulosa cell tumors. JCI insight, 8(22).

Ghaleb Y, et al. (2022) Whole Exome/Genome Sequencing Joint Analysis of a Family with Oligogenic Familial Hypercholesterolemia. Metabolites, 12(3).

Neitzel H, et al. (2022) Transmission ratio distortion of mutations in the master regulator of centriole biogenesis PLK4. Human genetics, 141(11), 1785.

Romanelli Tavares VL, et al. (2022) New locus underlying auriculocondylar syndrome (ARCND): 430 kb duplication involving TWIST1 regulatory elements. Journal of medical genetics, 59(9), 895.

Vona B, et al. (2021) A biallelic variant in CLRN2 causes non-syndromic hearing loss in humans. Human genetics, 140(6), 915.

Waseem SS, et al. (2021) A Homozygous AKNA Frameshift Variant Is Associated with Microcephaly in a Pakistani Family. Genes, 12(10).

Bezamat M, et al. (2021) Genome-wide family-based study in torus palatinus affected individuals. Archives of oral biology, 130, 105221.

Mol MO, et al. (2020) Clinical and pathologic phenotype of a large family with heterozygous STUB1 mutation. Neurology. Genetics, 6(3), e417.

Doll J, et al. (2020) Genetic Spectrum of Syndromic and Non-Syndromic Hearing Loss in Pakistani Families. Genes, 11(11).

Young KA, et al. (2019) Genome-Wide Association Study Identifies Loci for Liver Enzyme Concentrations in Mexican Americans: The GUARDIAN Consortium. Obesity (Silver Spring, Md.), 27(8), 1331.

Silva CT, et al. (2018) A combined linkage, microarray and exome analysis suggests MAP3K11 as a candidate gene for left ventricular hypertrophy. BMC medical genomics, 11(1), 22.

Bouhouche A, et al. (2018) Genetic Analysis of Undiagnosed Juvenile GM1-Gangliosidosis by Microarray and Exome Sequencing. *Case reports in genetics*, 2018, 8635698.

Cardamone G, et al. (2018) Genetic Association and Altered Gene Expression of CYBB in Multiple Sclerosis Patients. *Biomedicines*, 6(4).

Ta-Shma A, et al. (2018) Homozygous loss-of-function mutations in MNS1 cause laterality defects and likely male infertility. *PLoS genetics*, 14(8), e1007602.

Liaqat K, et al. (2018) Novel missense and 3'-UTR splice site variants in LHFPL5 cause autosomal recessive nonsyndromic hearing impairment. *Journal of human genetics*, 63(11), 1099.

Bourneuf E, et al. (2018) New susceptibility loci for cutaneous melanoma risk and progression revealed using a porcine model. *Oncotarget*, 9(45), 27682.

Eisenberger T, et al. (2018) A C-terminal nonsense mutation links PTPRQ with autosomal-dominant hearing loss, DFNA73. *Genetics in medicine : official journal of the American College of Medical Genetics*, 20(6), 614.

Liu S, et al. (2018) Retrospective Evaluation of Marker-Assisted Selection for Resistance to Bacterial Cold Water Disease in Three Generations of a Commercial Rainbow Trout Breeding Population. *Frontiers in genetics*, 9, 286.