

# Resource Summary Report

Generated by [dkNET](#) on Apr 26, 2025

## 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 resource, software application

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

**Funding:**

**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:** 20250421T053725+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

**Source:** [SciCrunch Registry](#)

---

## Usage and Citation Metrics

We found 75 mentions in open access literature.

**Listed below are recent publications.** The full list is available at [dkNET](#).

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).

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

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.

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.