## **Resource Summary Report**

Generated by <u>NIF</u> on May 20, 2025

# <u>GERP</u>

RRID:SCR\_000563 Type: Tool

**Proper Citation** 

GERP (RRID:SCR\_000563)

### **Resource Information**

URL: http://mendel.stanford.edu/SidowLab/downloads/gerp/

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**Description:** Software that identifies constrained elements in multiple alignments by quantifying substitution deficits. These deficits represent substitutions that would have occurred if the element were neutral DNA, but did not occur because the element has been under functional constraint. We refer to these deficits as Rejected Substitutions. Rejected substitutions are a natural measure of constraint that reflects the strength of past purifying selection on the element. GERP estimates constraint for each alignment column; elements are identified as excess aggregations of constrained columns. A false-positive rate (which is user-settable) is calculated using "shuffled" alignments in which the order of columns is randomized.

#### Abbreviations: GERP

**Synonyms:** Genomic Evolutionary Rate Profiling, GERP++, Genomic Evolutionary Rate Profiling: GERP, GERP2

Resource Type: software resource

Defining Citation: PMID:15965027, PMID:21152010

Keywords: genomic, evolution, rate profiling

Funding:

Resource Name: GERP

Resource ID: SCR\_000563

Alternate IDs: OMICS\_00174

Alternate URLs: https://sources.debian.org/src/gerp++/

**Record Creation Time:** 20220129T080202+0000

Record Last Update: 20250519T203056+0000

## **Ratings and Alerts**

No rating or validation information has been found for GERP.

No alerts have been found for GERP.

## Data and Source Information

Source: <u>SciCrunch Registry</u>

## **Usage and Citation Metrics**

We found 51 mentions in open access literature.

Listed below are recent publications. The full list is available at <u>NIF</u>.

Zhang X, et al. (2024) Clinical phenotype and genetic characteristics of SZT2 related diseases: A case report and literature review. Seizure, 114, 111.

Zhang X, et al. (2023) A novel heterozygous ATP1A2 pathogenic variant in a Chinese child with MELAS-like alternating hemiplegia. Molecular genetics & genomic medicine, 11(5), e2146.

Liu J, et al. (2023) Loss-of-function variants in KCTD19 cause non-obstructive azoospermia in humans. iScience, 26(7), 107193.

Gomes NL, et al. (2022) Contribution of Clinical and Genetic Approaches for Diagnosing 209 Index Cases With 46,XY Differences of Sex Development. The Journal of clinical endocrinology and metabolism, 107(5), e1797.

Barbosa P, et al. (2022) Computational prediction of human deep intronic variation. GigaScience, 12.

Wegscheid ML, et al. (2021) Patient-derived iPSC-cerebral organoid modeling of the 17q11.2 microdeletion syndrome establishes CRLF3 as a critical regulator of neurogenesis. Cell reports, 36(1), 109315.

Hecker N, et al. (2020) A genome alignment of 120 mammals highlights ultraconserved element variability and placenta-associated enhancers. GigaScience, 9(1).

Yang L, et al. (2020) Novel and de novo point and large microdeletion mutation in PRRT2related epilepsy. Brain and behavior, 10(5), e01597.

Yang L, et al. (2019) Novel and de novo mutation of PCDH19 in Girls Clustering Epilepsy. Brain and behavior, 9(12), e01455.

Osipova E, et al. (2019) RepeatFiller newly identifies megabases of aligning repetitive sequences and improves annotations of conserved non-exonic elements. GigaScience, 8(11).

Wang P, et al. (2018) Association of IL17RC and COL6A1 genetic polymorphisms with susceptibility to ossification of the thoracic posterior longitudinal ligament in Chinese patients. Journal of orthopaedic surgery and research, 13(1), 109.

Schrauwen I, et al. (2018) Novel digenic inheritance of PCDH15 and USH1G underlies profound non-syndromic hearing impairment. BMC medical genetics, 19(1), 122.

Flood VH, et al. (2018) Common VWF sequence variants associated with higher VWF and FVIII are less frequent in subjects diagnosed with type 1 VWD. Research and practice in thrombosis and haemostasis, 2(2), 390.

Wang M, et al. (2018) DeFine: deep convolutional neural networks accurately quantify intensities of transcription factor-DNA binding and facilitate evaluation of functional non-coding variants. Nucleic acids research, 46(11), e69.

Dadaev T, et al. (2018) Fine-mapping of prostate cancer susceptibility loci in a large metaanalysis identifies candidate causal variants. Nature communications, 9(1), 2256.

Roscito JG, et al. (2018) The genome of the tegu lizard Salvator merianae: combining Illumina, PacBio, and optical mapping data to generate a highly contiguous assembly. GigaScience, 7(12).

Koch E, et al. (2017) A Temporal Perspective on the Interplay of Demography and Selection on Deleterious Variation in Humans. G3 (Bethesda, Md.), 7(3), 1027.

Zheng WS, et al. (2017) EP300 contributes to high-altitude adaptation in Tibetans by regulating nitric oxide production. Zoological research, 38(3), 163.

Li Q, et al. (2017) InterVar: Clinical Interpretation of Genetic Variants by the 2015 ACMG-AMP Guidelines. American journal of human genetics, 100(2), 267.

Zhang Y, et al. (2017) Accurate and reproducible functional maps in 127 human cell types

via 2D genome segmentation. Nucleic acids research, 45(17), 9823.