Resource Summary Report

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PhenoTips

RRID:SCR_006340
Type: Tool

Proper Citation

PhenoTips (RRID:SCR_006340)

Resource Information

URL: http://phenotips.cs.toronto.edu/

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Description: A software tool providing a Web interface and a database back-end for collecting clinical symptoms and physical findings observed in patients with genetic disorders. The main goals of this software are * To allow for collecting patient data in standard formats, enabling effortless data exchange and automated search in annotated gene and disease databases, and * To provide advanced functionalities and a friendly user interface that help reduce the clinician""s workload, permitting seamless use of this application within the clinician""s routine. PhenoTips uses the Human Phenotype Ontology (HPO) to express clinical phenotypes, and provides a friendly interface with error-tolerant, predictive search of phenotypic descriptions. PhenoTips closely mirrors clinician workflows: observations can be recorded directly during the patient encounter, and the interface is compatible with any device that runs a modern Web browser. The clinician can record demographic information, family history, medical history, various standard measurements, phenotypic abnormalities detected in the patient, pertinent indications that were not observed and that can be helpful for differential diagnosis, relevant images depicting manifestations of the patient""s disorders, and additional notes for each of these categories. The software automatically plots growth curves, selects phenotypes reflecting abnormal measurements, instantly finds OMIM disorders matching the phenotypic description and suggests other symptoms to investigate in order to reach a more accurate diagnosis.

Abbreviations: PhenoTips

Synonyms: PhenoTips: phenotyping made easy

Resource Type: software application, software resource

Keywords: clinical symptom, physical finding, clinical, phenotype, demographic information, family history, medical history, standard measurement, indication, image, note, growth curve

Related Condition: Genetic disorder

Availability: Free

Resource Name: PhenoTips

Resource ID: SCR_006340

Alternate IDs: nlx_152049

Record Creation Time: 20220129T080235+0000

Record Last Update: 20260727T040411+0000

Ratings and Alerts

No rating or validation information has been found for PhenoTips.

No alerts have been found for PhenoTips.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 24 mentions in open access literature.

Listed below are recent publications. The full list is available at nidm-terms .

Hou H, et al. (2025) Assessing the diagnostic impact of blood transcriptome profiling in a pediatric cohort previously assessed by genome sequencing. NPJ genomic medicine, 10(1), 51.

Krumb E, et al. (2024) Proactive systematic hemophilia carrier screening: a step toward gender equity in hemophilia care. Blood advances, 8(20), 5268.

Wallis M, et al. (2024) Experience of the first adult-focussed undiagnosed disease program in Australia (AHA-UDP): solving rare and puzzling genetic disorders is ageless. Orphanet journal of rare diseases, 19(1), 288.

van der Sanden BPGH, et al. (2023) The performance of genome sequencing as a first-tier test for neurodevelopmental disorders. European journal of human genetics : EJHG, 31(1), 81.

Driver HG, et al. (2022) Genomics4RD: An integrated platform to share Canadian deep-phenotype and multiomic data for international rare disease gene discovery. Human mutation, 43(6), 800.

Yates J, et al. (2021) Finding commonalities in rare diseases through the undiagnosed diseases network. Journal of the American Medical Informatics Association : JAMIA,

28(8), 1694.

Parikh JR, et al. (2021) A data-driven architecture using natural language processing to improve phenotyping efficiency and accelerate genetic diagnoses of rare disorders. HGG advances, 2(3).

Amburgey K, et al. (2021) A Cross-Sectional Study of Nemaline Myopathy. Neurology, 96(10), e1425.

Almowil ZA, et al. (2021) Concept libraries for automatic electronic health record based phenotyping: A review. International journal of population data science, 6(1), 1362.

Salvatore M, et al. (2020) Improving diagnosis for rare diseases: the experience of the Italian undiagnosed Rare diseases network. Italian journal of pediatrics, 46(1), 130.

Reuter MS, et al. (2020) The Cardiac Genome Clinic: implementing genome sequencing in pediatric heart disease. Genetics in medicine : official journal of the American College of Medical Genetics, 22(6), 1015.

Urreizti R, et al. (2020) Five new cases of syndromic intellectual disability due to KAT6A mutations: widening the molecular and clinical spectrum. Orphanet journal of rare diseases, 15(1), 44.

Yaramis A, et al. (2020) COL4A1-related autosomal recessive encephalopathy in 2 Turkish children. Neurology. Genetics, 6(1), e392.

Costain G, et al. (2020) Genome Sequencing as a Diagnostic Test in Children With Unexplained Medical Complexity. JAMA network open, 3(9), e2018109.

Mroczek M, et al. (2020) Four Individuals with a Homozygous Mutation in Exon 1f of the PLEC Gene and Associated Myasthenic Features. Genes, 11(7).

Töpf A, et al. (2020) Sequential targeted exome sequencing of 1001 patients affected by unexplained limb-girdle weakness. Genetics in medicine : official journal of the American College of Medical Genetics, 22(9), 1478.

López-Martín E, et al. (2018) SpainUDP: The Spanish Undiagnosed Rare Diseases Program. International journal of environmental research and public health, 15(8).

Lionel AC, et al. (2018) Improved diagnostic yield compared with targeted gene sequencing panels suggests a role for whole-genome sequencing as a first-tier genetic test. Genetics in medicine : official journal of the American College of Medical Genetics, 20(4), 435.

Jia J, et al. (2018) RDAD: A Machine Learning System to Support Phenotype-Based Rare Disease Diagnosis. Frontiers in genetics, 9, 587.

Mullegama SV, et al. (2017) Coupling clinical exome sequencing with functional characterization studies to diagnose a patient with familial Mediterranean fever and MED13L haploinsufficiency syndromes. Clinical case reports, 5(6), 833.