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Ústav zdravotnických informací a statistiky ČR
Institute of Health Information and Statistics of the Czech Republic



UNIVERZITA
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Human Phenotype Ontology

Česká terminologie pro popis fenotypu,
přínosy a možnosti využití

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1. Ústav biologie a lékařské genetiky 2. LF UK a FNM

2. Ústav zdravotnických informací a statistiky

Human Phenotype Ontology

- Standardizovaná terminologie pro popis **fenotypu**
- Fenotyp:
 - Všechny pozorovatelné a případně vyšetřitelné **znaky** a **vlastnosti** jedince (pacienta).
 - Genotyp + prostředí + epigenetické faktory
- **Ontologie:**
 - Explicitní specifikace konceptualizace
 - Orientovaný acyklický graf
- Vychází z katalogu OMIM,
- **13 000 termínů**
- 156 000 anotací k dědičným nemocem
- Další relace: geny, MeSH, PubMed, UMLS, SNOMED CT...
- Licence: volné užití, citace, změny jen vývojáři HPO
- <https://github.com/obophenotype/human-phenotype-ontology>

The screenshot shows the HPO website homepage. At the top is a dark teal navigation bar with the HPO logo and menu items: About, Data, Tools, Resources, and Community. Below this is a large teal banner featuring the HPO logo and the text "human phenotype ontology". A search bar is present with a dropdown menu set to "All" and the placeholder text "Search for phenotypes, diseases or genes...". Below the search bar, there are examples: "e.g. Arachnodactyly | Marfan syndrome | FBN1".

The Human Phenotype Ontology

The Human Phenotype Ontology (HPO) provides a standardized vocabulary of phenotypic abnormalities encountered in human disease. Each term in the HPO describes a phenotypic abnormality, such as [Atrial septal defect](#). The HPO is currently being developed using the medical literature, Orphanet, DECIPHER, and OMIM. HPO currently contains over 13,000 terms and over 156,000 annotations to hereditary diseases. The HPO project and others have developed software for phenotype-driven differential diagnostics, genomic diagnostics, and translational research. The HPO is a flagship product of the [Monarch Initiative](#), an NIH-supported international consortium dedicated to semantic integration of biomedical and model organism data with the ultimate goal of improving biomedical research. The HPO, as a part of the Monarch Initiative, is a central component of one of the [13 driver projects](#) in the [Global Alliance for Genomics and Health \(GA4GH\) strategic roadmap](#).

[Learn More About HPO](#)

News & Updates

- [June 2023 HPO release & updates](#) June 6, 2023
- [April 2023 HPO release & updates](#) April 6, 2023
- [June 2022 HPO release](#) June 12, 2022

[View All News](#)

Exomiser
Evaluate variants based on the predicted pathogenicity.

Phenomizer
Rank disease differential diagnosis by clinical features.

Příklad termínu

- **Název:** Hypertelorism
- **Identifikátor:** HP:0000316
- **Definice:** Interpupillary distance more than 2 SD above the mean (alternatively, the appearance of an increased interpupillary distance or widely spaced eyes).
- **Synonyma:**
 - Excessive orbital separation
 - Increased distance between eye sockets
 - Increased distance between eyes
 - Increased interpupillary distance
 - Ocular hypertelorism
 - Wide-set eyes
 - Widely spaced eyes
 - Widened interpupillary distance


Hypertelorism HP:0000316

Interpupillary distance more than 2 SD above the mean (alternatively, the appearance of an increased interpupillary distance or widely spaced eyes).

Synonyms: Excessive orbital separation, Increased distance between eye sockets, Increased distance between eyes, Increased interpupillary distance, Ocular hypertelorism, Wide-set eyes, Widely spaced eyes, Widened interpupillary distance

Pubmed References: [PMID:19125427](#)

Cross References: MSH:D006972, SNOMEDCT_US:194021007, SNOMEDCT_US:22006008, UMLS:C0020534

 [Export Associations](#)

Disease Associations		
Disease Id	Disease Name	Associated Genes
ORPHA:276413	10q22.3q23.3 microdeletion syndrome	
ORPHA:94063	12q14 microdeletion syndrome	HMGA2 [8091] LEMD3 [23592]
ORPHA:261120	14q11.2 microdeletion syndrome	
ORPHA:264200	14q22q23 microdeletion syndrome	
ORPHA:401935	14q24.1q24.3 microdeletion syndrome	
ORPHA:314585	15q overgrowth syndrome	

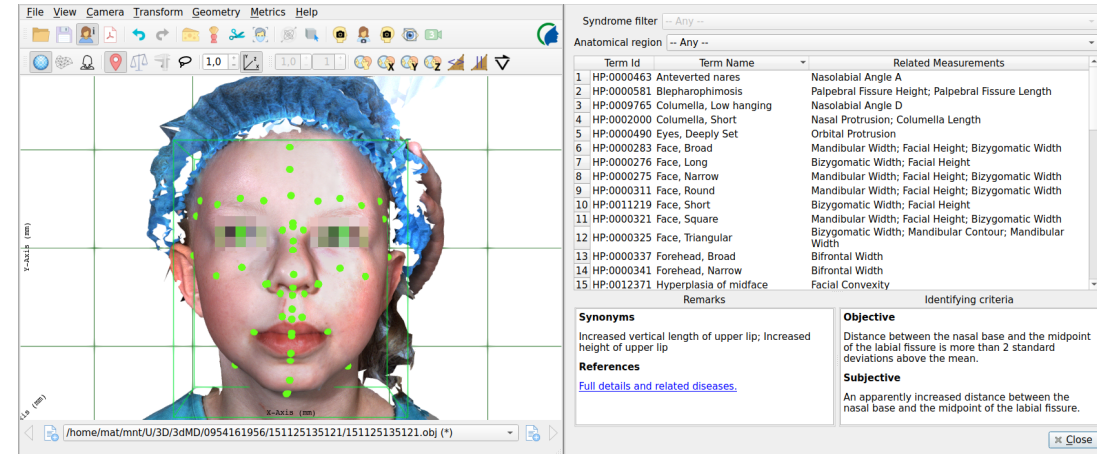
Displaying 20 out of 1167. [View all](#)

Gene Associations	
Gene Symbol	Associated Diseases
ABCD4 [5826]	(OMIM:614857) - Methylmalonic aciduria and homocystinuria, Cblj type
ACOX1 [51]	(OMIM:264470) - Peroxisomal acyl-CoA oxidase deficiency (ORPHA:2971) - Peroxisomal acyl-CoA oxidase deficiency (OMIM:618960) - Mitchell syndrome
ACTA1 [58]	(ORPHA:171433) - Intermediate nemaline myopathy (ORPHA:171436) - Typical nemaline myopathy (OMIM:161800) - Nemaline myopathy 3 (ORPHA:171430) - Severe congenital nemaline myopathy (ORPHA:97240) - Zebra body myopathy (ORPHA:171439) - Childhood-onset nemaline myopathy (OMIM:616852) - Myopathy, scapulohumeroperoneal (ORPHA:97244) - Rigid spine syndrome (ORPHA:2020) - Congenital fiber-type disproportion myopathy

Displaying 20 out of 760. [View all](#)

Využití HPO

- Možnost určení podobnosti fenotypu
- Informační systémy, registry
- Bioinformatická analýza dat se sekvenováním nové generace
 - filtrování a/nebo prioritizace nalezených variant
- Návrhy diagnózy na základě zadaných příznaků
 - Phenomizer, Face2Gene
- Systémy navrhuující HPO termíny z fotografií či 3D modelů
- Text mining
- „Match making“



The screenshot displays a 3D facial model software interface. The central part shows a 3D model of a child's face with green dots indicating measurement points. The interface includes a menu bar (File, View, Camera, Transform, Geometry, Metrics, Help) and a toolbar. On the right, there is a panel with a table of HPO terms and related measurements.

Term Id	Term Name	Related Measurements
1 HP-0000463	Anteverted nares	Nasolabial Angle A
2 HP-0000581	Blepharophimosis	Palpebral Fissure Height; Palpebral Fissure Length
3 HP-0009765	Columella, Low hanging	Nasolabial Angle D
4 HP-0002000	Columella, Short	Nasal Protusion; Columella Length
5 HP-0000490	Eyes, Deeply Set	Orbital Protusion
6 HP-0000283	Face, Broad	Mandibular Width; Facial Height; Bizygomatic Width
7 HP-0000276	Face, Long	Bizygomatic Width; Facial Height
8 HP-0000275	Face, Narrow	Mandibular Width; Facial Height; Bizygomatic Width
9 HP-0000311	Face, Round	Mandibular Width; Facial Height; Bizygomatic Width
10 HP-0011219	Face, Short	Bizygomatic Width; Facial Height
11 HP-0000321	Face, Square	Mandibular Width; Facial Height; Bizygomatic Width
12 HP-0000325	Face, Triangular	Bizygomatic Width; Mandibular Contour; Mandibular Width
13 HP-0000337	Forehead, Broad	Bifrontal Width
14 HP-0000341	Forehead, Narrow	Bifrontal Width
15 HP-0012371	Hyperplasia of midface	Facial Convexity

Below the table, there are sections for Synonyms, References, Objective, and Subjective. The Objective section states: "Distance between the nasal base and the midpoint of the labial fissure is more than 2 standard deviations above the mean." The Subjective section states: "An apparently increased distance between the nasal base and the midpoint of the labial fissure."

Překlad HPO do češtiny

- Implementace v českých IS a expertních systémech
- Širší možnosti použití terminologie odbornou veřejností (starší lékaři, SZP)
- Možnosti strojového zpracování nestructurovaných zpráv v češtině
- První překlady od roku 2016, od 2018 další překladatelé, od roku 2020 intenzivněji
- 437 172 slov
- Online překladatelská platforma Crowdin
- Aktuální stav: přeloženo 100 %, probíhá revize termínů přidaných v posledních dvou letech

Search languages

AZ



★ Czech

100% · 76%



Dutch

10% · 0%



Italian

9% · 0%



Japanese

9% · 0%



Norwegian

0% · 0%



Portuguese

0% · 0%

PY

Russian

9% · 0%



Turkish

78% · 49%

← CZECH > HPO LABELS, SYNONYMS, ...

- ☰ "abnormality of the immune system" ✕
- Anti-H1 antibody positivity
- The presence of autoantibodies in the blood circulation that r...
- Anti-H4 antibody positivity
- The presence of autoantibodies in the blood circulation that r...
- Anti-H3 antibody positivity
- The presence of autoantibodies in the blood circulation that r...
- Anti-H2A antibody positivity
- The presence of autoantibodies in the blood circulation that r...
- Anti-H2B antibody positivity
- The presence of autoantibodies in the blood circulation that r...
- Absent peripheral lymph nodes in presence of infection
- The absence of any palpable lymph nodes in the presence of ...
- Vulvar abscess
- A circumscribed area of pus or necrotic debris in the vulvar re...
- Inguinal abscess

SOURCE STRING

Inguinal abscess

CONTEXT ▾ REQUEST

Key: HP_0033590_label
HPO_classes
HP_0033590_label
label of HP:0033590 (Inguinal abscess) ; abnormality of the immune system

Inguinální absces

16 · 17 SAVE

CZECH TRANSLATIONS ▾

Inguinální absces
Marek Turnovec (turnovec) 2 months ago

TM AND MT SUGGESTIONS ▾

Intrarenální absces
InguinalIntrarenal abscess

COMMENTS

No comments yet

New Features

- you can now find an international version of HPO at <http://purl.obolibrary.org/obo/hp/hp-international.owl>. This will contain translations of many HPO phenotypes into french, czech, turkish and dutch. More languages are being added in future releases.
- The individual language profiles can be browsed at <https://github.com/obophenotype/human-phenotype-ontology/tree/master/src/ontology/translations>, and accessed using the persistent IRI prefix `http://purl.obolibrary.org/obo/hp/translations/`, for example: <http://purl.obolibrary.org/obo/hp/translations/hp-fr.babelon.tsv> to obtain the latest french language profile as a spreadsheet. We do not currently provided official PURLs for the versioned language profiles, but they can be accessed using regular GitHub versioning like this: <https://github.com/obophenotype/human-phenotype-ontology/blob/v2023-04-05/src/ontology/translations/hp-fr.babelon.tsv>.
- The French translation of HPO is carried out by INSERM, US14- Orphanet, with funding from the French Ministry of Health, Direction Générale de la Santé, in the framework of the French National Plan for Rare Diseases.

Překladačský tým – poděkování

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