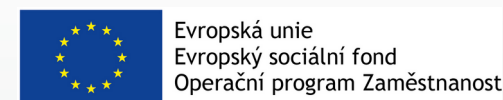


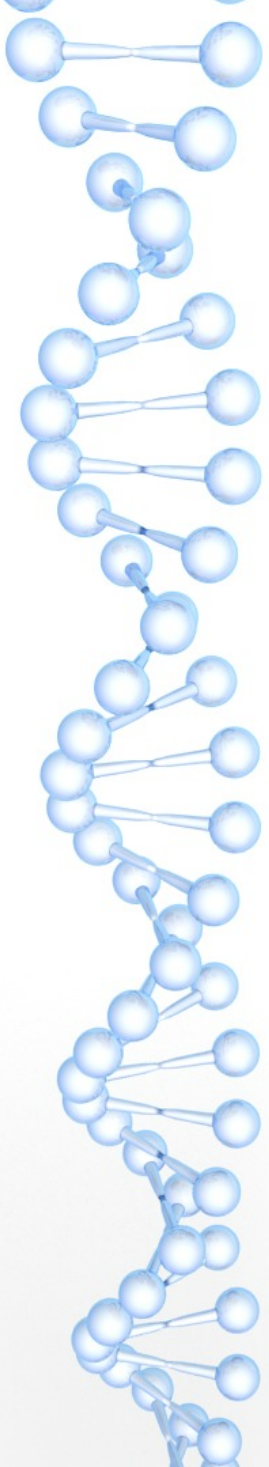


Překlad Human Phenotype Ontology do češtiny

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Osnova

- Co je HPO?
- K čemu je to dobré?
- Proč překládat?
- Kdo překládá?
- Jak překládáme?



Co je to HPO?

- Fenotyp:
 - Všechny pozorovatelné a případně vyšetřitelné znaky a vlastnosti jedince (pacienta).
 - Genotyp + prostředí + epigenetika = **fenotyp**.
- Ontologie:
 - „explicitní specifikace konceptualizace“
- **Standardizovaná terminologie** pro popis fenotypu.



Hypertelorismus

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

<https://hpo.jax.org/>



All

Search for phenotypes, diseases or genes...

e.g. [Arachnodactyly](#) | [Marfan syndrome](#) | [FBN1](#)


Hypertelorism HP:0000316

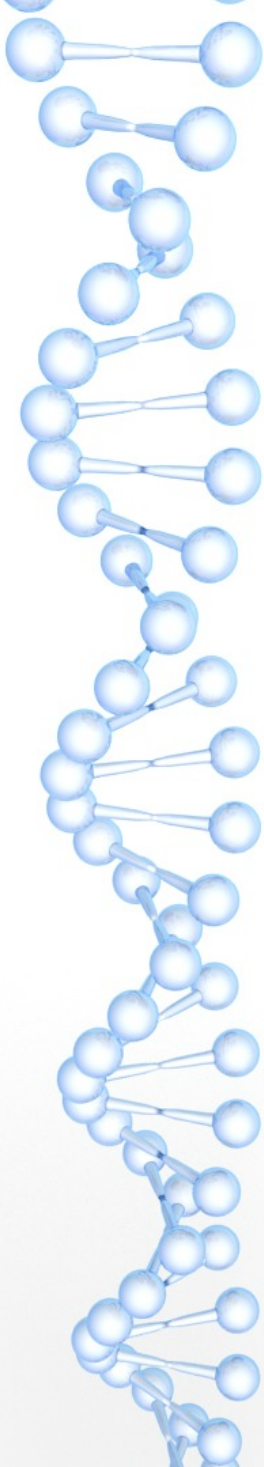
Interpupillary distance more than 2 SD above the mean (alternatively, the appearance of an increased interpupillary distance or widely spaced eyes).

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

PubMed References: [PMID:19125427](#)

Cross References: [MSH:D006972](#), [SNOMEDCT_US:194021007](#), [UMLS:C0020534](#), [SNOMEDCT_US:22006008](#)

 Export Associations



Disease Associations		Gene Associations
Filter by disease		
Disease Id	Disease Name	Associated Genes
ORPHA:284984	Aneurysm-osteoarthritis Syndrome	SMAD3 [4088]
ORPHA:1104	Anophthalmia Plus Syndrome	
ORPHA:83	Antley-bixler Syndrome	FGFR2 [2263]
OMIM:201750	Antley-bixler Syndrome With Genital Anomalies And Disordered Steroidogenesis	POR [5447]
ORPHA:87	Apert Syndrome	FGFR2 [2263]
OMIM:101200	Apert Syndrome	FGFR2 [2263]

Items per page: 51 – 100 of 1051 < >

Disease Associations		Gene Associations
Filter by gene		
Gene Symbol	Associated Diseases	
CHD3 [1107]	(ORPHA:564) - Meckel Syndrome (OMIM:618205) - Snijders Blok-campeau Syndrome	
CHD4 [1108]	(OMIM:617159) - Sifrim-hitz-weiss Syndrome	
CHD7 [55636]	(OMIM:612370) - Hypogonadotropic Hypogonadism 5 With Or Without Anosmia (ORPHA:478) - Kallmann Syndrome (ORPHA:432) - Normosmic Congenital Hypogonadotropic Hypogonadism (OMIM:214800) - Charge Syndrome (ORPHA:39041) - Omenn Syndrome (ORPHA:138) - Charge Syndrome	
CHD8 [57680]	(OMIM:615032) - Autism, Susceptibility To, 18	

Items per page: 51 – 100 of 634 < >



K čemu je to dobré?

- Bioinformatická analýza dat získaných ze sekvenování nové generace (NGS)
 - Exomiser
 - Genomiser
- Návrhy diagnózy na základě zadaných příznaků
 - Phenomizer
- Standardizovaný popis pacientů pro „match making“

Proč překládat?

- Širší možnosti použití terminologie (starší lékaři, SZP)
- Možnosti strojového zpracování nestructurovaných zpráv v češtině



★ Czech

12% · 0%



Dutch

14% · 0%



French

25% · 0%



German

100% · 4%



Italian

12% · 0%



Japanese

11% · 0%



Portuguese

1% · 0%



Russian

11% · 0%

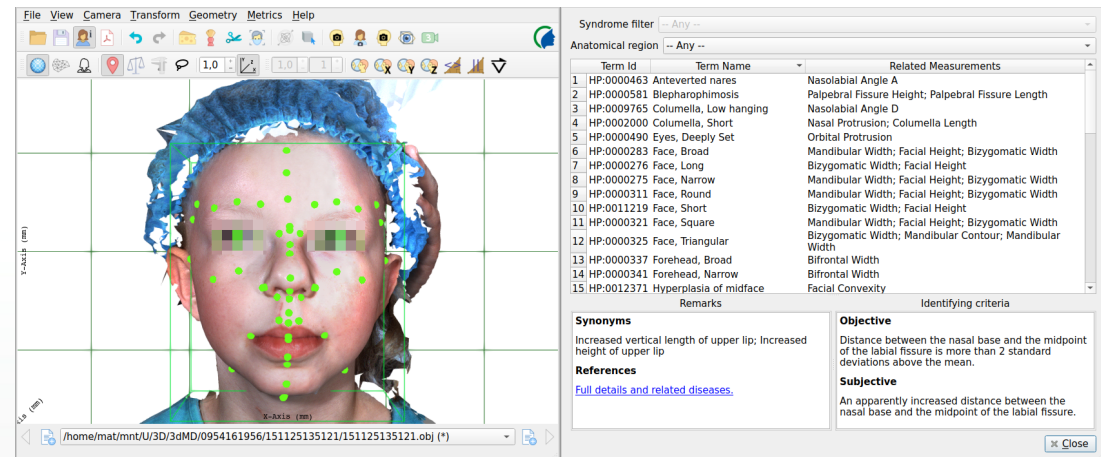
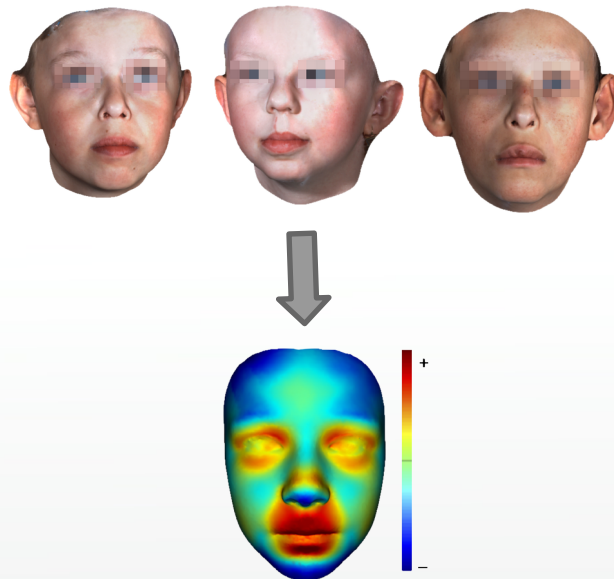


Turkish

100% · 71%

Kdo překládá?

- Tým lékařů – klinických genetiků z Ústavu biologie a lékařské genetiky 2. LF UK a FNM.
- Část týmu překládala terminologii vzácných onemocnění
- Zkušenosti s popisem dysmorfických pacientů a digitální fenotypizací (3D skenování)



Jak překládáme

- Online platforma Crowdin

The screenshot shows the Crowdin project dashboard for 'HPO translation'. At the top, there are navigation tabs for 'Home', 'Activity', 'Reports', 'Discussions', and 'Tasks'. Below this, a 'Translations' section displays progress bars for various languages: Czech (12% · 0%), Dutch (14% · 0%), French (25% · 0%), German (100% · 4%), Italian (12% · 0%), Japanese (11% · 0%), Portuguese (1% · 0%), Russian (11% · 0%), and Turkish (100% · 71%). On the right side, there is a 'Description' section for the 'HPO Translation Project', which explains the project's goal of translating HPO labels and synonyms into various languages. It also includes contact information for Sebastian Köhler (drseb) and a 'Show More' button. Below the description, there is a 'Details' section with project statistics: ID: 172282, Source language: English, Project members: 104, Words to translate: 335 192, Created: 5 years ago, and Last activity: 4 minutes ago. At the bottom, a 'Managers' section lists Sebastian Köhler (drseb) as the owner with a 'Contact' button.

The screenshot shows the Crowdin translation editor interface for the term 'Hypertelorismus'. The top navigation bar indicates the current project is 'CZECH' and the specific task is 'HPO LABELS, SYNONYMS, ...'. The main editing area shows the source string 'Hypertelorismus' and its context. Below the source string, there are tabs for 'Strings', 'Context', 'Translations', and 'Everything'. The 'Translations' tab is active, showing a list of translations for the source string. The first translation is 'Hypertelorismus' by Marek Turnovec (turnovec) 3 years ago. Below this, there are 'TM AND MT SUGGESTIONS' from Watson (IBM) Translator and Crowdin Translate. The interface also includes a 'SAVE' button and a '13 · 15' indicator.

Díky za pozornost



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