New Application: Integrate PEDIA!

Prioritization of Exome Data by Image Analysis

What is PEDIA?

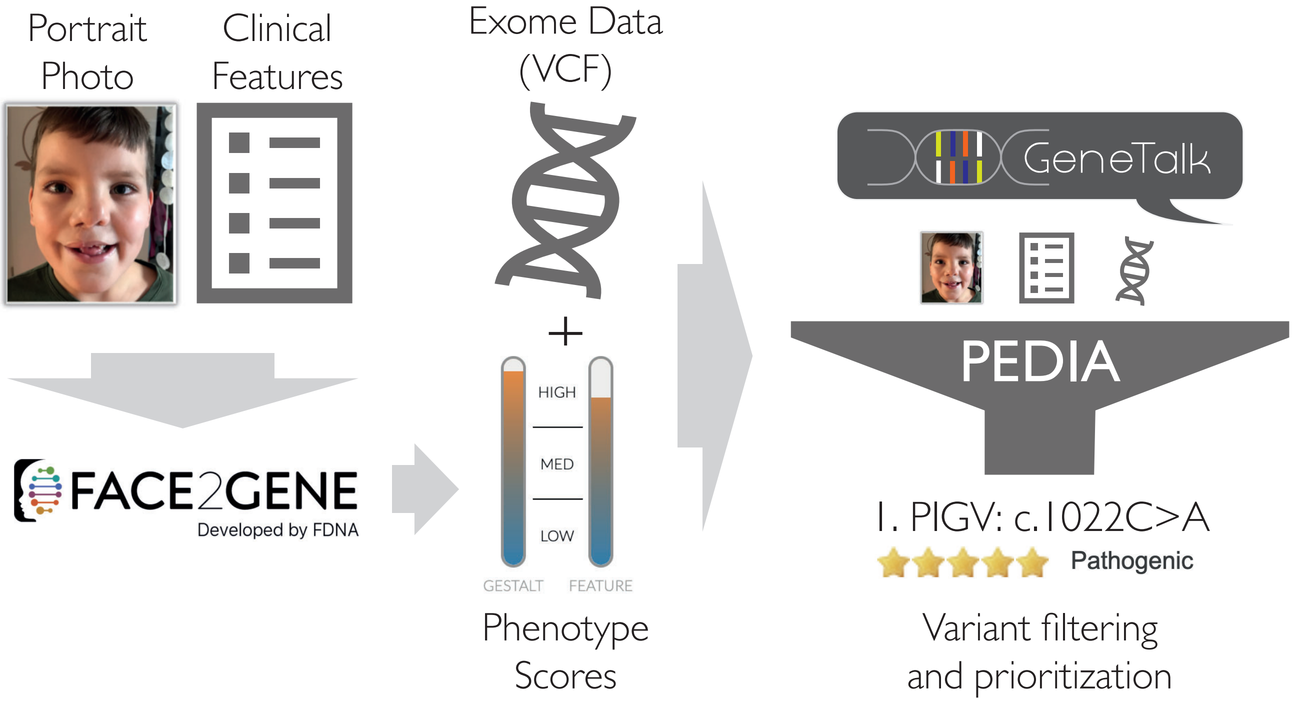
PEDIA is an acronym for **P**rioritization of **E**xome **D**ata by **I**mage **A**nalysis. PEDIA combines information from the phenotypic and molecular level for sequence variant interpretation. PEDIA does not only work with the clinical features encoded in **H**uman **P**henotype **O**ntology (**HPO**) terminology, it harnesses the full information of a human syndromic face with artificial intelligence. PEDIA is built on **n**ext **g**eneration of **p**henotyping technology (**NGP**) from **Face2Gene** developed by FDNA: The convolutional neural network **DeepGestalt** is able to detect dysmorphic features in frontal photographs of patients (*Gurovich* et al., Nature Medicine 2019). The comparison of the gestalt to phenotypic models of more than 300 syndromes results in similarity scores that can be mapped to genes and used in variant interpretation.

The PEDIA classifier is constantly updated and every solved exome case improves its performance. Currently PEDIA is trained on more than 650 extensively studied and molecularly confirmed monogenic syndromic cases. With PEDIA scoring the pathogenic mutation is at the top position in nearly nine out of ten cases (*Hsieh et al.*, Genetics in Medicine 2019).

What are the requirements?

In order to make full use of the possibilities of the PEDIA-Score, information from all three domains should be available:

- Portrait Photo
- Clinical Features
- Exome Data


Portrait Photo
Clinical Features
Exome Data (VCF)
FACE2GENE
Developed by FDNA
+

Phenotype Scores

1. PIGV: c.1022C>A
★★★★★ Pathogenic
Variant filtering and prioritization

analyze human sequence variants

www.gene-talk.de

GeneTalk

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How to ingerate PEDIA scoring into your exome analysis pipeline

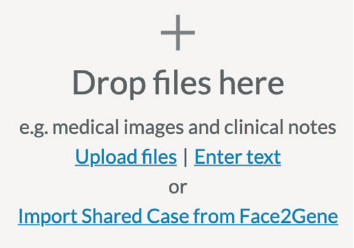
GeneTalk is the first provider to fully integrate NGP scores from DeepGestalt into its exome analysis framework. However, there are many ways to adapt the PEDIA approach to your needs. What is your use case? Do you have the laboratory inhouse? How does your current IT infrastructure look like? Who has access to which kind of data during the analysis? Which molecular scores do you like to work with? We would like to find a solution that fits your requirements.

On **www.gene-talk.de** a new widget will be launched - the **Phenobot**. Once you have uploaded your patient's genetic data, you will find the following button:

LAUNCH PHENOBOT

In the pop-up window you can upload the portrait photo of the patient under **Collect** (A). DeepGestalt is started automatically. The results of the phenotypic analysis can be found under **Analyze**. Here you can also enter the phenotypic characteristics of your patient in HPO terms (B). Soon also the tool Text2Phenotype (T2P) will be integrated. T2P is a tool that uses natural language processing (NLP) to extract HPO terms from the notes of the clinical encounter. Please make sure which of the suggested terms are present or not. If you already uploaded the patient‘s data in **Face2Gene CLINIC** you can import the data directly via **Import Shared Case** (A). A list of your patients in your Face2Gene account will be provided (C).

A



Drop files here

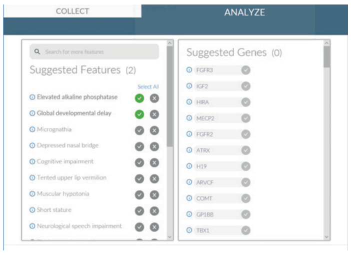
e.g. medical images and clinical notes

Upload files | Enter text

or

Import Shared Case from Face2Gene

B



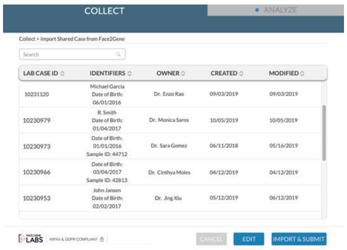
Suggested Features (2)

- Elevated alkaline phosphatase
- Global developmental delay
- Micrognathia
- Depressed nasal bridge
- Cognitive impairment
- Tented upper lip vermilion
- Muscular hypotonia
- Short stature
- Neurological speech impairment

Suggested Genes (0)

- FGFR3
- IGF2
- HIRA
- MECP2
- FGFR2
- ATRX
- H19
- ARVCF
- COMT
- GP1BB
- TBX1

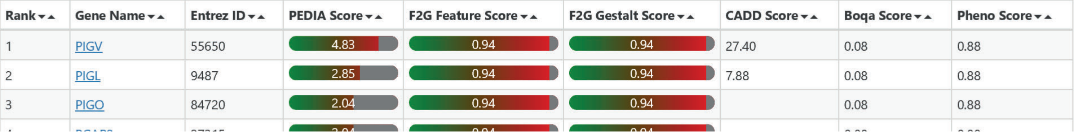
C



Collect > Import Shared Case from Face2Gene

LAB CASE ID	IDENTIFIERS	OWNER	CREATED	MODIFIED
10231120	Michael Garcia Date of Birth: 06/01/2016	Dr. Enzo Rao	09/03/2019	09/03/2019
10230979	R. Smith Date of Birth: 01/04/2017	Dr. Monica Saros	10/05/2019	10/05/2019
10230973	Date of Birth: 01/01/2016 Sample ID: 44712	Dr. Sara Gomez	06/11/2018	05/16/2019
10230966	Date of Birth: 03/04/2017 Sample ID: 42813	Dr. Cinthya Moles	04/12/2019	04/12/2019
10230953	John Jansen Date of Birth: 02/02/2017	Dr. Jing Xiu	05/12/2019	06/12/2019

After you collected all phenotypic data, DeepGestalt does its magic. As soon as you close the pop-up session, all NGP scores are transferred and the PEDIA analysis starts. With PEDIA we aim to provide you with the best prioritization score on the gene level, however, in the VCF viewer you can add any additional score that you desire and sort accordingly.



Rank ▾▴	Gene Name ▾▴	Entrez ID ▾▴	PEDIA Score ▾▴	F2G Feature Score ▾▴	F2G Gestalt Score ▾▴	CADD Score ▾▴	Boqa Score ▾▴	Pheno Score ▾▴
1	PIGV	55650	4.83	0.94	0.94	27.40	0.08	0.88
2	PIGL	9487	2.85	0.94	0.94	7.88	0.08	0.88
3	PIGO	84720	2.04	0.94	0.94		0.08	0.88

If you are interested in using next-generation phenotyping scores from Face2Gene in your own analysis pipeline, GeneTalk would be happy to assist you with the integration. Please get in contact with us, so that we can learn more about the special needs of your IT infrastructure.