What is PEDIA?

PEDIA is an acronym for Prioritization of Exome Data by Image Analysis. PEDIA combines information from the phenotypic and molecular level for sequence variant interpretation. PEDIA does not only work with the clinical features encoded in Human Phenotype Ontology (HPO) terminology, it harnesses the full information of a human syndromic face with artificial intelligence. PEDIA is built on next generation of phenotyping 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

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<th>Portrait Photo</th>
<th>Clinical Features</th>
<th>Exome Data (VCF)</th>
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**FACE2GENE**
Developed by FDNA

**PEDIA**

1. PIGV: c.1022C>A

Phenotype Scores

Variant filtering and prioritization

www.gene-talk.de
How to integrate 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](http://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](image)

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).

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.

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.