FACE2GENE
Smart Phenotyping. Better Genetics.

FACE2GENE CONNECT
Face2Gene.com/LabsAPI

What is Face2Gene?

Face2Gene is a suite of phenotyping apps that facilitate comprehensive and precise genetic evaluations. Face2Gene increases the confidence of clinicians and bioinformatics by prioritizing syndromes and variants in clinic and in lab.

What is Face2Gene Connect™?

Face2Gene Connect™ gives developers the ability to access a patient's de-identified phenotypic data through a secure API. Adopting institutions can develop their own proprietary applications that leverage Face2Gene's unique gestalt analysis in combination with annotated clinical data, while streamlining and simplifying the workload for ordering clinicians.

What does Face2Gene Connect™ provide?

- Enables clinicians to provide clinical details to a laboratory within Face2Gene, making it easier to order genetic testing
- Obtain a detailed phenotype report with your patient's detected and annotated HPO features
- Review a short list of plausible syndromes with OMIM IDs
- Sift through a list of the most clinically relevant genes

How can Face2Gene Connect™ be helpful in genetic testing interpretation?

- Face2Gene Connect™ provides laboratory teams with a detailed phenotype report to aid in variant analysis by filtering and prioritizing variants based on clinical annotations of features, syndrome differentials and scoring of syndromes and variants using proprietary feature and gestalt analysis.
- A few examples of how Face2Gene supports rare disease evaluation:
    - Face2Gene analysis has been shown to assist in whole exome sequencing interpretation (Gripp KW, et al. 2016; Mensah MA, et al. 2016 [abstract])
    - Face2Gene may also provide evidence supporting a syndrome in cases with non-typical clinical presentation (Mensah MA, et al. 2016 [abstract])
    - Deep phenotyping provided by Face2Gene analysis could be key in identifying syndromic forms of autism and could help in the selection of diagnostic molecular tests (Varga NA 2016 [abstract]).
    - The detection rate of Face2Gene for Cornelia de Lange syndrome was found to be comparable to dysmorphology experts (Basel-Vanagaite, L 2016)

What does a lab agree to when using Face2Gene Connect™?

- Face2Gene Connect™ is open, flexible, and free of charge to bioinformatics companies and genetic testing labs
- We encourage approved entities to use the Face2Gene Connect™ logo in their applications, websites, and marketing materials (including their requisition orders), to inform their clients that Face2Gene Connect™ is enabled
- Use of our insignia is subject to our Terms of Use

Patient Privacy

Face2Gene uses advanced technologies to protect patient information.

- No personal health information (PHI) from patients is transmitted via Face2Gene Connect™
- All patient photos uploaded by our clinical users are converted into a de-identified mathematical facial descriptor. This data set is used for the Face2Gene analysis, while the original photo is encrypted and stored on separate disk volume, accessible only to our clinical users and other healthcare providers whom they actively approve. See data sharing policy for more information.
- Compliant with HIPAA and European Union (EU) privacy rules and standards
- Your clients do not need to change their standard patient consent forms

EU
HIPAA & EU DATA PRIVACY COMPLIANT

FACE2GENE CONNECT

Your Face2Gene Connect™ API

Below you will find your Lab Key code which is required for access.

ACCOUNT (INSTITUTION NAME)

LAB KEY

YOUR NAME

EMAIL

Retrospective Case Examples

The following are test cases that have been analyzed and supplied for your review by Dr. Peter Krawitz of Charité (Berlin) and GeneTalk. These assess a combination of gestalt-match, feature-match, and variant-scores for disease-gene prediction.

Sample Face2Gene Case URL Format

https://app.face2gene.com//lab-api/case-info?lab_key={Lab_Key}&case_code={Case_Code}

CASE 1

35424.NA18570. Achondroplasia Syndrome

- Face2Gene Phenotype
- Filtered Exome VCF

CASE 2

44698.HG02137.Kabuki Syndrome

- Face2Gene Phenotype
- Filtered Exome VCF

CASE 3

41339.HG00190.BWS Syndrome

- Face2Gene Phenotype
- Filtered Exome VCF

CASE 4

35305.NA12829. Alagille Syndrome

- Face2Gene Phenotype
- Filtered Exome VCF

API Methodology

1 Case-info: Obtain case information

Request

Method	URL
GET	https://app.face2gene.com/lab-api/case-info

Type	Params	Values
HEAD	lab_key	string
HEAD	case_code	string

lab_key must be sent with all client requests. The api_key helps the server to validate the request source.
case_code determines which case info will be retrieved (should be supplied by the face2gene user)

Response

Status	Response

200	{
	"gender": <gender>
	"ethnicity_array": <ethnicity>,
	"inheritance_array": <inheritance mode>,
	"detectedSyndromes": <detected syndromes>,
	"detectedFeatures": <detected features>,
	"selectedSyndromes": <selected syndromes>,
	"selectedFeatures": <selected features>,
	"genomic": <genomic data>
	}

404	"no case_code provided or case_code is in an incorrect format"
404	"The page you are trying to view does not exist"

2 vcf-case-info: Get the uploaded vcf file of a case

Request

Method	URL
POST	https://app.face2gene.com/lab-api/vcf-case-info

Type	Params	Values
HEAD	lab_key	string
HEAD	case_code	string

lab_key must be sent with all client requests. The api_key helps the server to validate the request source.
case_code determines which case info will be retrieved (should be supplied by the face2gene user)

Response

Status	Response
200	<VCF file>
404	"The page you are trying to view does not exist"
404	"no vcf file for case 44441"

FACE2GENE CONNECT

Example Request

https://app.face2gene.com/lab-api/case-info?lab_key=abcdef12345&case_code=ABC123:

Example Response


```
{
    "gender": "Male",
    "ethnicity_array": ["Caucasian"],
    "inheritance_array": ["Autosomal recessive"],
    "detectedSyndromes": [{
        "syndrome_name": "Hennekam Lymphangiec-
tasia-Lymphedema
Syndrome 1; HKLLS1",
        "omim_id": "235510",
        "gestalt_match_score": "3.55337",
        "feature_match_score": "0.88",
        "genes": [{
            "gene_omim_id": "612753",
            "gene_symbol": "CCBE1"
        }]
    }, {
        "syndrome_name": "Apert Syndrome",
        "omim_id": "101200",
        "score": "1.80513",
        "genes": [{
            "gene_omim_id": "176943",
            "gene_symbol": "FGFR2"
        }]
    }, {
        "syndrome_name": "Kleefstra Syndrome",
        "omim_id": "610253",
        "gestalt_match_score": "3.55337",
        "feature_match_score": "0.88",
        "genes": [{
            "gene_omim_id": "607001",
            "gene_symbol": "EHMT1"
        }]
    }, {
        "syndrome_name": "Mucopolysaccharidoses",
        "omim_id": "607014",
        "score": "1.56236",
        "genes": [{
            "gene_omim_id": "252800",
            "gene_symbol": "IDUA"
        }, {
            "gene_omim_id": "605270",
            "gene_symbol": "SGSH"
        }, {
            "gene_omim_id": "609701",
            "gene_symbol": "NAGLU"
        }, {
            "gene_omim_id": "611542",
            "gene_symbol": "ARSB"
        }, {
            "gene_omim_id": "611499",
            "gene_symbol": "GUSB"
        }, {
            "gene_omim_id": "300823",
            "gene_symbol": "IDS"
        }, {
            "gene_omim_id": "607071",
            "gene_symbol": "HYAL1"
        }, {
            "gene_omim_id": "610453",
            "gene_symbol": "HGSNAT"
        }, {
            "gene_omim_id": "612222",
            "gene_symbol": "GALNS"
        }, {
            "gene_omim_id": "611458",
            "gene_symbol": "GLB1"
        }]
    }, {
        "syndrome_name": "Charge Syndrome",
        "omim_id": "214800",
        "gestalt_match_score": "3.55337",
        "feature_match_score": "0.88",
        "genes": [{
            "gene_omim_id": "608166",
            "gene_symbol": "SEMA3E"
        }, {
            "gene_omim_id": "608892",
            "gene_symbol": "CHD7"
        }]
    }, {
        "syndrome_name": "3MC Syndrome",
        "omim_id": "257920",
        "score": "1.18386",
        "genes": [{
            "gene_omim_id": "600521",
            "gene_symbol": "MASP1"
        }, {
            "gene_omim_id": "612502",
            "gene_symbol": "COLEC11"
        }]
    }, {
        "syndrome_name": "Kaufman Oculocerebro-
facial Syndrome;
KOS",
        "omim_id": "244450",
        "gestalt_match_score": "3.55337",
        "feature_match_score": "0.88",
        "genes": [{
            "gene_omim_id": "608047",
            "gene_symbol": "UBE3B"
        }]
    }, {
        "syndrome_name": "Cri-Du-Chat Syndrome",
        "omim_id": "123450",
        "gestalt_match_score": "3.55337",
        "feature_match_score": "0.88",
        "genes": []
    }, {
        "syndrome_name": "Arthrogryposis, Distal,
Type 2A; DA2A",
        "omim_id": "193700",
        "gestalt_match_score": "3.55337",
        "feature_match_score": "0.88",
        "genes": [{
            "gene_omim_id": "160720",
            "gene_symbol": "MYH3"
        }]
    }, {
        "syndrome_name": "Opitz GBBB Syndrome,
Type II; GBBB2",
        "omim_id": "145410",
        "gestalt_match_score": "3.55337",
        "feature_match_score": "0.88",
        "genes": [{
            "gene_omim_id": "614140",
            "gene_symbol": "SPECC1L"
        }]
    }],
    "detectedFeatures": [{
        "feature_name": "Thick upper lip vermilion",
        "hpo": "HP:0000215",
        "status": "Present"
    }, {
        "feature_name": "Micrognathia",
        "hpo": "HP:0000347",
        "status": "Present"
    }, {
        "feature_name": "Hypertelorism",
        "hpo": "HP:0000316",
        "status": "Present"
    }],
    "selectedSyndromes": [{
        "syndrome_name": "Hennekam Lymphangiecta-
sia-Lymphedema
Syndrome 1; HKLLS1",
        "omim_id": "235510",
        "genes": [{
            "gene_omim_id": "612753",
            "gene_symbol": "CCBE1"
        }]
    }],
    "selectedFeatures": [{
        "feature_name": "Downslanted palpebral fissures",
        "hpo": "HP:0000494",
        "status": "Present"
    }, {
        "feature_name": "Breast hypertrophy",
        "hpo": "HP:0010313",
        "status": "Present"
    }, {
        "feature_name": "Abnormality of the coagulation
cascade",
        "hpo": "HP:0003256",
        "status": "Present"
    }, {
        "feature_name": "Abnormality of the breast",
        "hpo": "HP:0000769",
        "status": "Present"
    }],
    "genomic": [{
        "test": [{
            "value": "Monogenic",
            "key": "Mutation Type"
        }, {
            "value": "Specific mutation testing",
            "key": "Molecular Test"
        }, {
            "value": "AADACL3",
            "key": "Gene Name"
        }, {
            "value": "Heterozygous",
            "key": "Genotype"
        }, {
            "value": "Protein Level",
            "key": "Notation"
        }],
        "description": [{
            "key": "Inheritance Mode",
            "value": "Autosomal Recessive"
        }, {
            "key": "result",
            "value": "Normal"
        }]
    }]
}
```