Imaging applications in dysmorphology and syndromology

Hilde Peeters

2024 February 13. Dysmorphology and Syndromology

Clinical dysmorphology

1960-70

DS delineation at the molecular level

1990

Micro array

Reverse dysmorphology

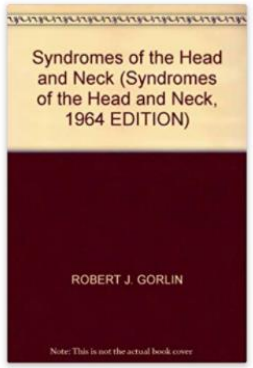
2006

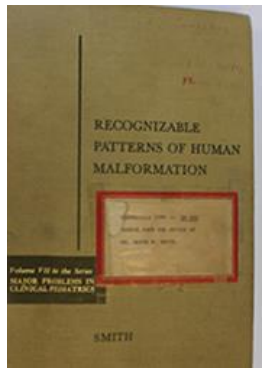
NGS

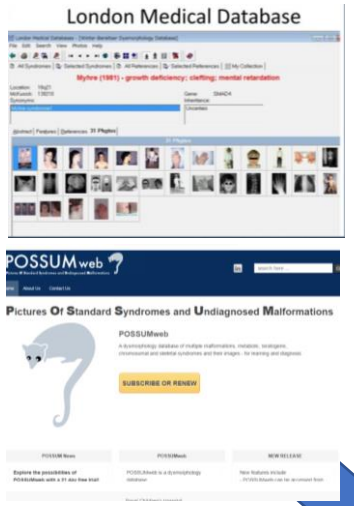
Reverse dysmorphology

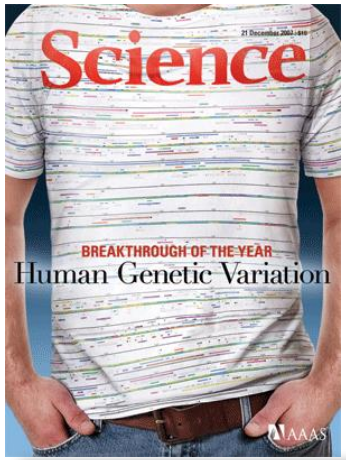
2012

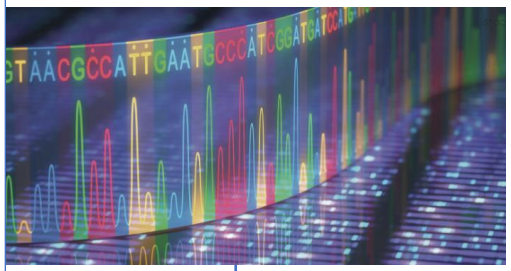
2024





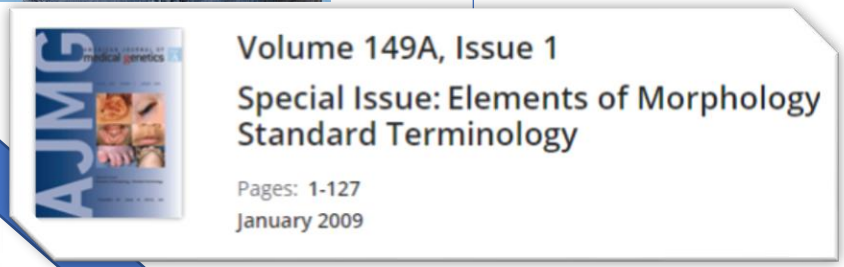




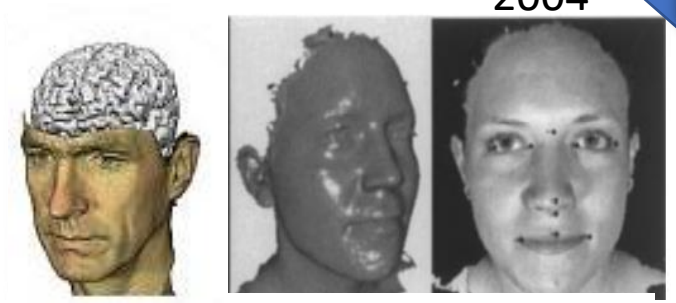


syndrome

phenotype



2004



3D Analysis of Facial Morphology

Peter Hammond,[1]* Tim J. Hutton,[1] Judith E. Allanson,[2] Linda E. Campbell,[3] Raoul C.M. Hennekam,[4] Sean Holden,[5] Michael A. Patton,[6] Adam Shaw,[6] I. Karen Temple,[7] Matthew Trotter,[8] Kieran C. Murphy,[9] and Robin M. Winter[10]

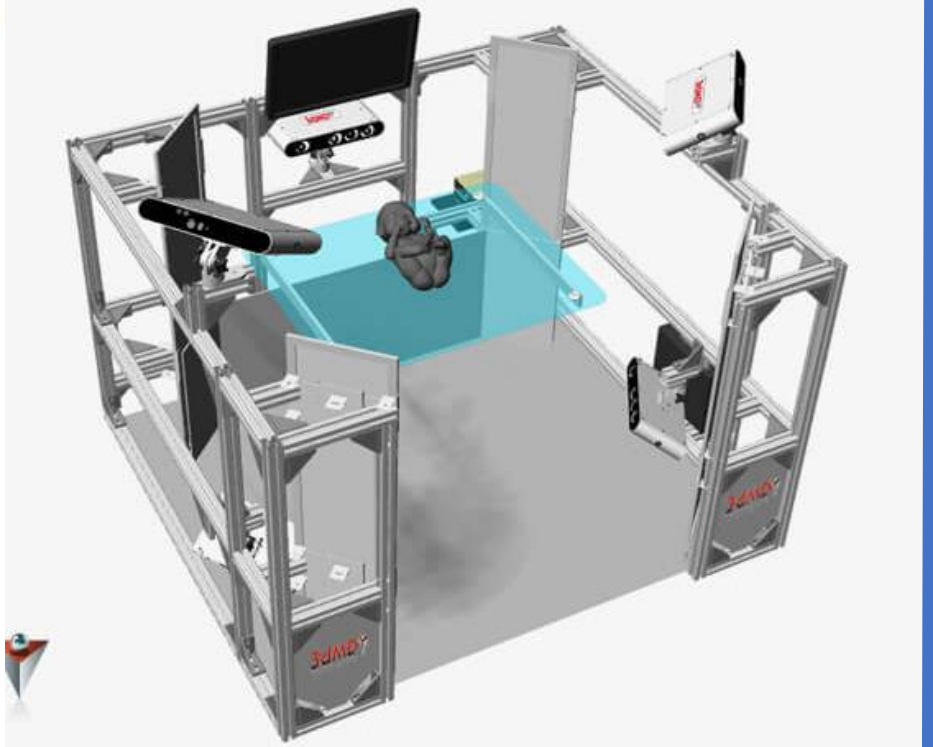
2011

FDNA

3D facial images of 7057 subjects: 3327 with 396 different syndromes, 727 of their relatives, and 3003 unrelated, unaffected subjects.
Hallgrimsson et al. 2020

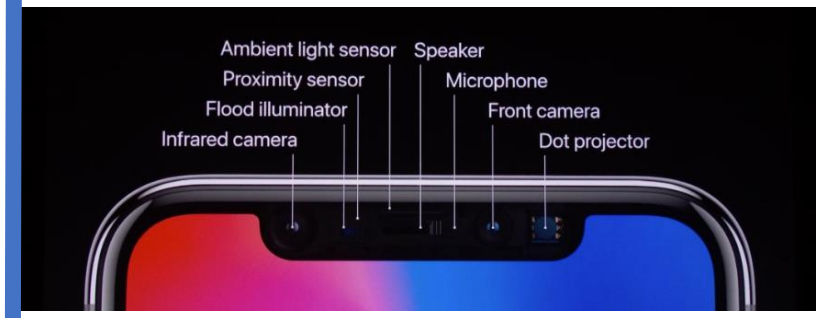
~2000 3D images rare developmental disorders .
P Hammond

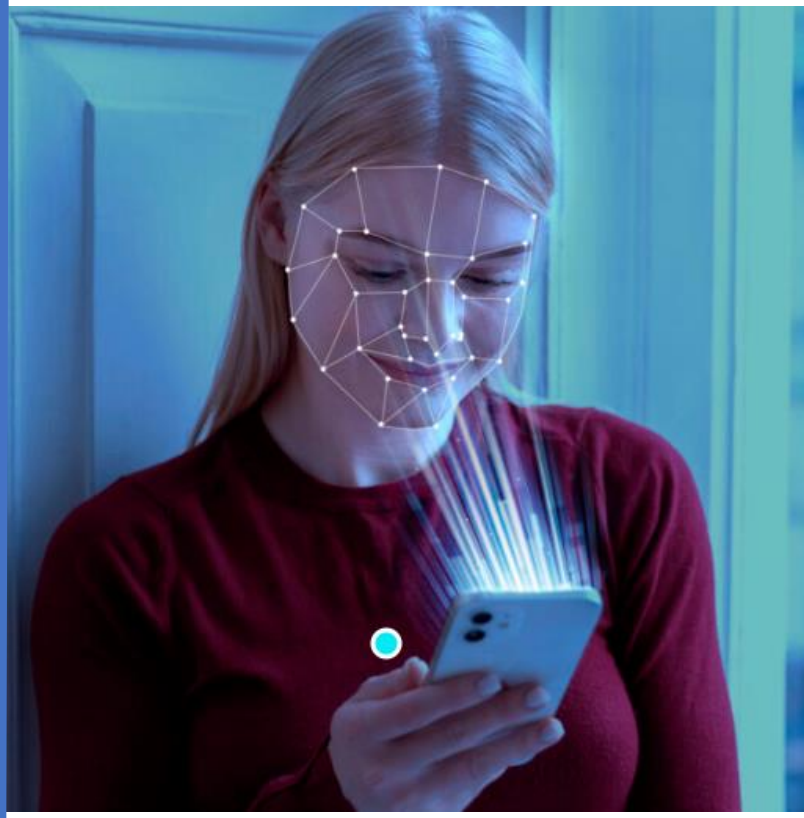
3DMD



Vectra H1

True Depth Camera Apple





Applications of 2D and 3D imaging

- Syndrome recognition, classification, ranking
- Explore the phenotypic variability of syndromic conditions
    - Monogenic conditions: study the relation of the facial phenotype with molecular diagnosis
    - Expand on the phenotypic spectrum -> milder phenotype
    - Variant interpretation
- Normal versus abnormal facial variation

Applications of 2D and 3D imaging

- Syndrome recognition, classification, ranking

Micro array

NGS

Clinical dysmorphology

DS delineation at the molecular level

Reverse dysmorphology

Reverse dysmorphology

1960-70

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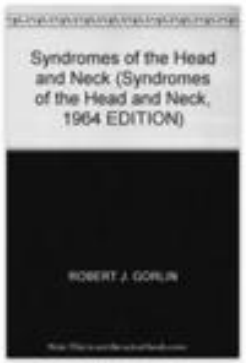
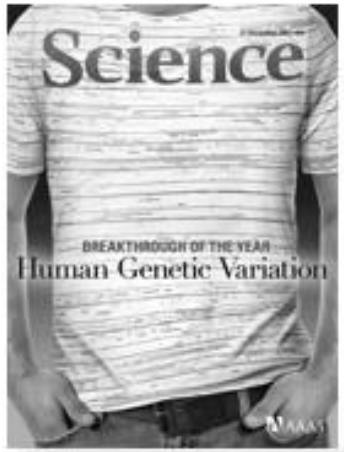
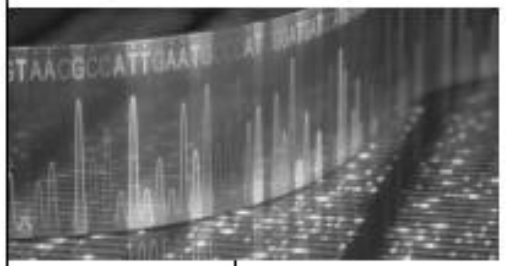
2021







syndrome

phenotype

Volume 149A, Issue 1

Special Issue: Elements of Morphology Standard Terminology

Pages: 1-127

January 2009

2011

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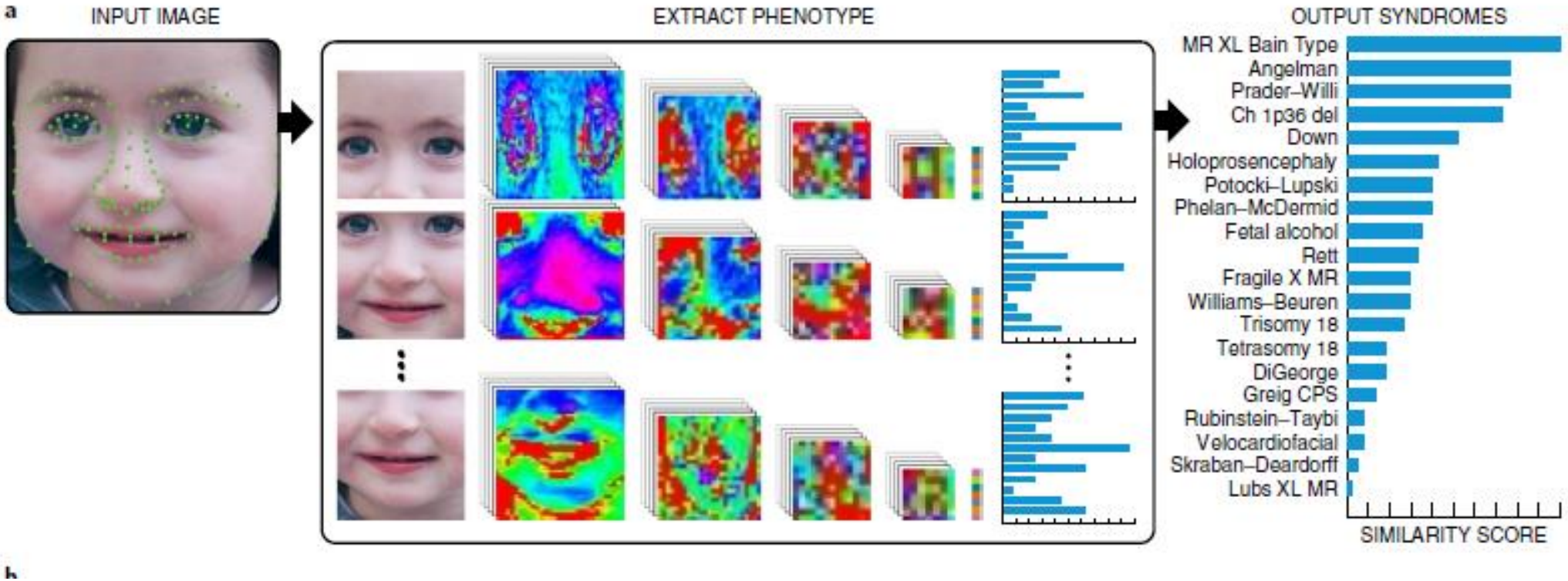
3D Analysis of Facial Morphology

FACE2GENE CLINIC FORUMS LIBRARY RESEARCH

CLINIC | CASE LIST | NEW CASE | SUPPORT | Prof. Hilde Peeters



Picture from FDNA

Received: 4 November 2019 | Revised: 20 February 2020 | Accepted: 20 April 2020

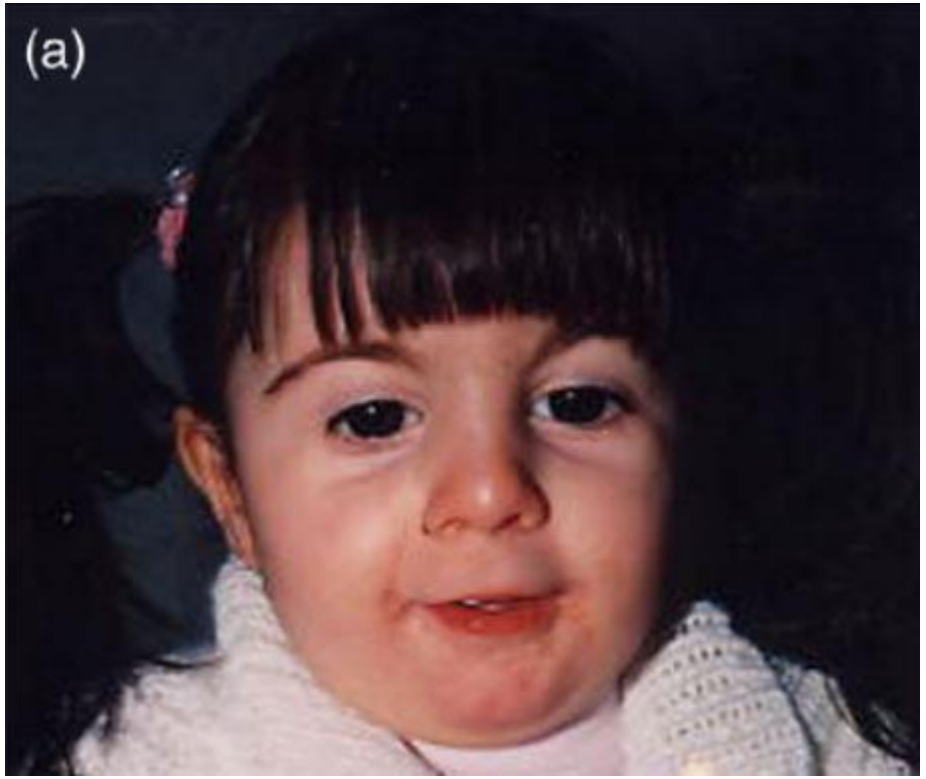
DOI: 10.1002/ajmg.a.61611

ORIGINAL ARTICLE

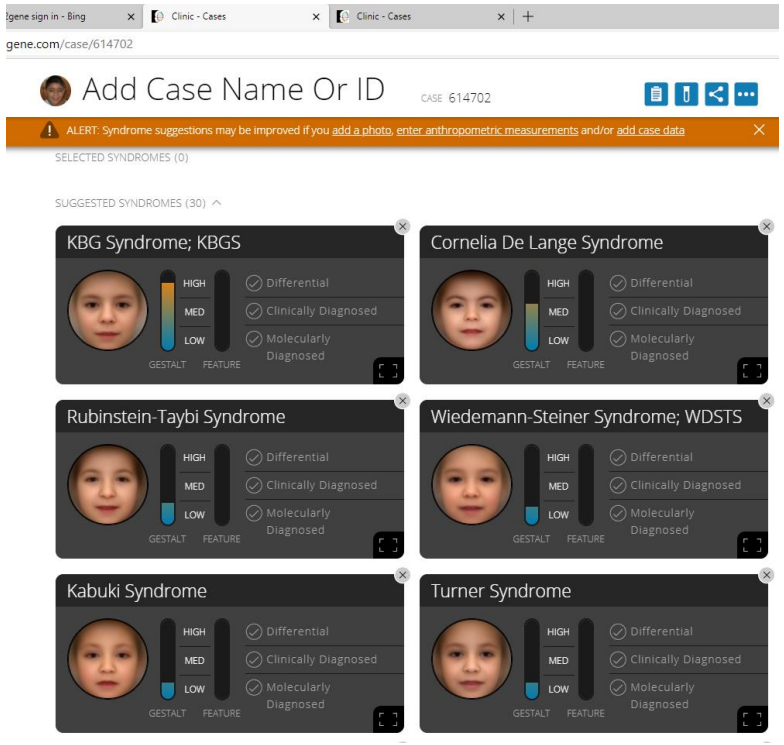
AMERICAN JOURNAL OF medical genetics PART A WILEY

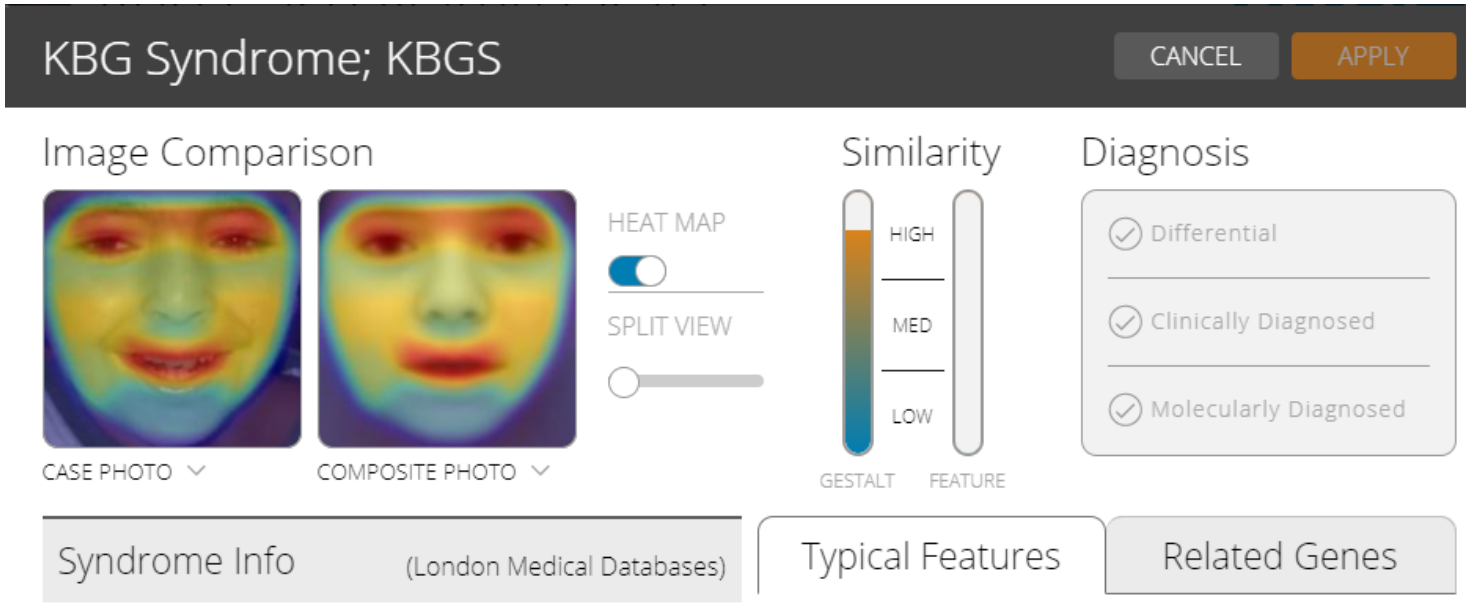
Pathogenic variants in *EP300* and *ANKRD11* in patients with phenotypes overlapping Cornelia de Lange syndrome

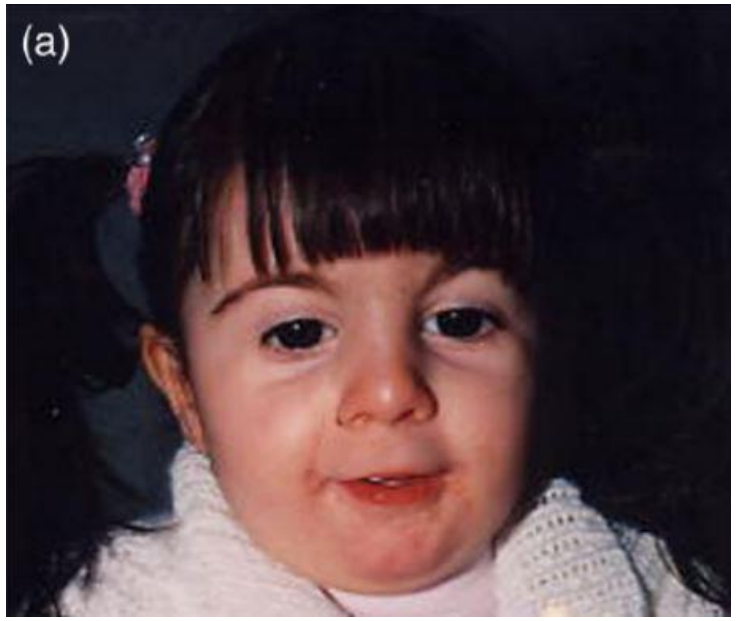
Francesco Cucco[1] | Patrizia Sarogni[1] | Sara Rossato[2] | Mirella Alpa[3] | Alessandra Patimo[1] | Ana Latorre[4] | Cinzia Magnani[5] | Beatriz Puisac[4] | Feliciano J. Ramos[4] | Juan Pié[4] | Antonio Musio[1]

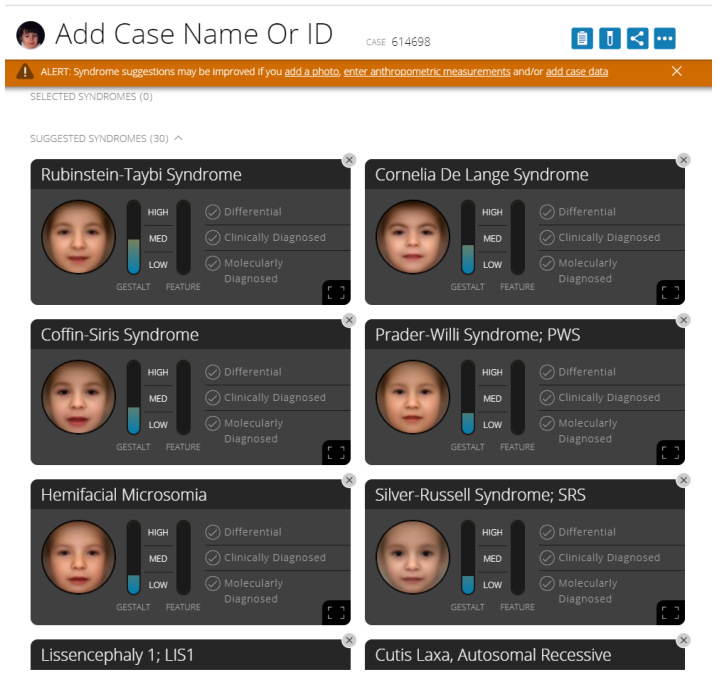


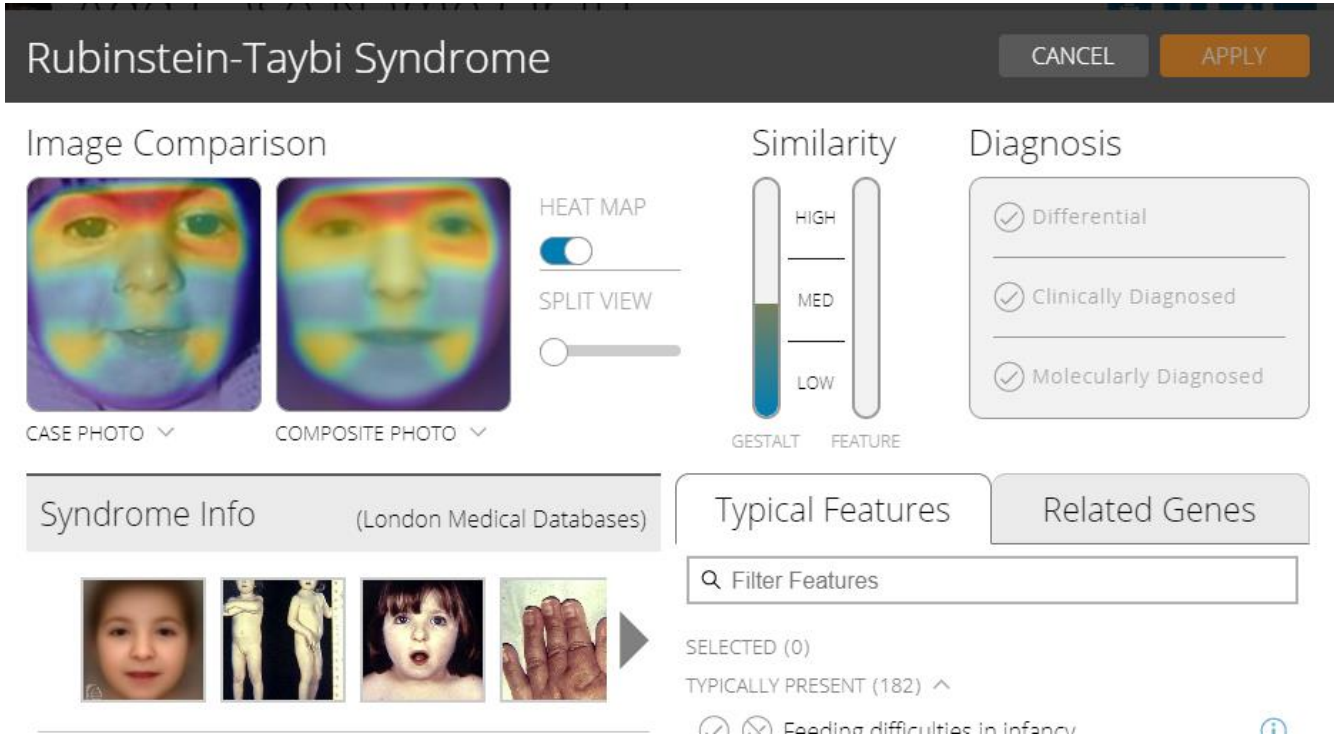

(b)


gene.com/case/614702
Add Case Name Or ID
CASE 614702
ALERT: Syndrome suggestions may be improved if you add a photo, enter anthropometric measurements and/or add case data
SELECTED SYNDROMES (0)
SUGGESTED SYNDROMES (30)
KBG Syndrome; KBGS
Cornelia De Lange Syndrome
Rubinstein-Taybi Syndrome
Wiedemann-Steiner Syndrome; WDSTS
Kabuki Syndrome
Turner Syndrome
HIGH
MED
LOW
GESTALT
FEATURE
Differential
Clinically Diagnosed
Molecularly Diagnosed


KBG Syndrome; KBGS
CANCEL
APPLY
Image Comparison
HEAT MAP
SPLIT VIEW
CASE PHOTO
COMPOSITE PHOTO
Similarity
HIGH
MED
LOW
GESTALT
FEATURE
Diagnosis
Differential
Clinically Diagnosed
Molecularly Diagnosed
Syndrome Info
(London Medical Databases)
Typical Features
Related Genes


(a)


Add Case Name Or ID
CASE 614698
ALERT: Syndrome suggestions may be improved if you add a photo, enter anthropometric measurements and/or add case data
SELECTED SYNDROMES (0)
SUGGESTED SYNDROMES (30)
Rubinstein-Taybi Syndrome
Cornelia De Lange Syndrome
Coffin-Siris Syndrome
Prader-Willi Syndrome; PWS
Hemifacial Microsomia
Silver-Russell Syndrome; SRS
Lissencephaly 1; LIS1
Cutis Laxa, Autosomal Recessive
HIGH
MED
LOW
GESTALT
FEATURE
Differential
Clinically Diagnosed
Molecularly Diagnosed


Rubinstein-Taybi Syndrome
CANCEL
APPLY
Image Comparison
HEAT MAP
SPLIT VIEW
CASE PHOTO
COMPOSITE PHOTO
Similarity
HIGH
MED
LOW
GESTALT
FEATURE
Diagnosis
Differential
Clinically Diagnosed
Molecularly Diagnosed
Syndrome Info
(London Medical Databases)
Typical Features
Related Genes
Filter Features
SELECTED (0)
TYPICALLY PRESENT (182)

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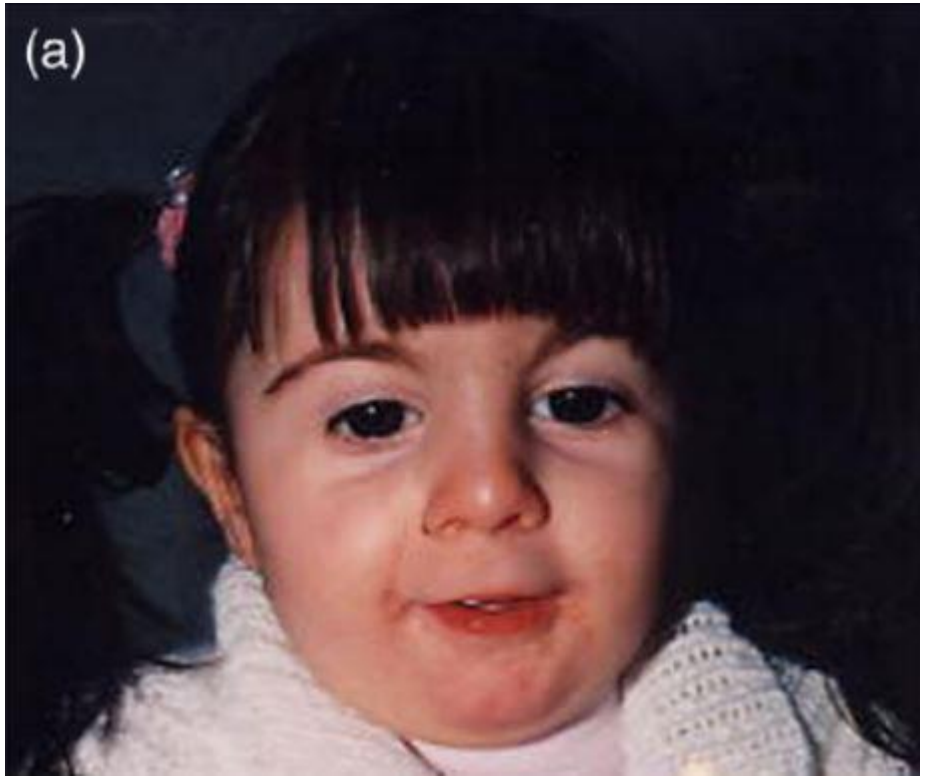
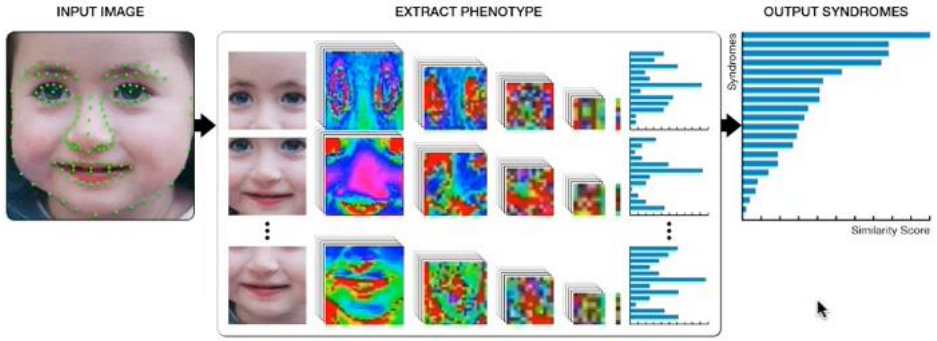
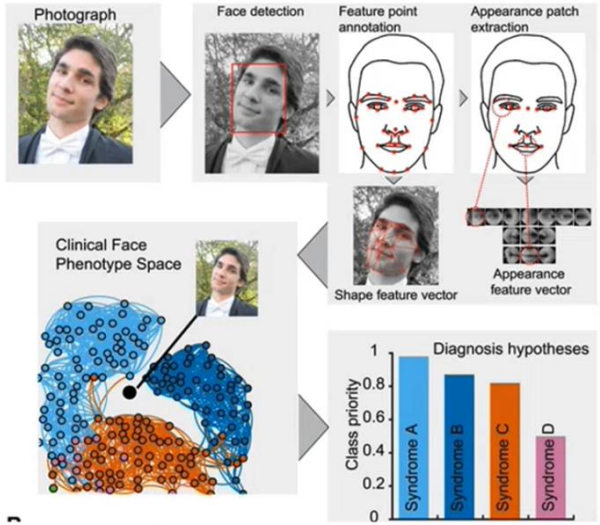


Part based syndrome classification

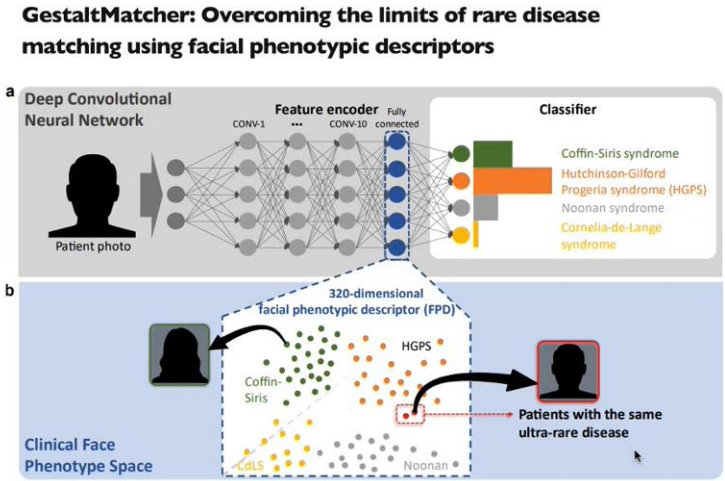


Y. Gurovich et al., "Identifying facial phenotypes of genetic disorders using deep learning," Nat. Med., vol. 25, no. 1, Art. no. 1, Jan. 2019, doi: 10.1038/s41591-018-0279-0

Clinical phenotype face space



Q. Ferry et al., "Diagnostically relevant facial gestalt information from ordinary photos," eLife, vol. 3, p. e02020, Jun. 2014, doi: 10.7554/eLife.02020.



T.-C. Hsieh et al., "GestaltMatcher: Overcoming the limits of rare disease matching using facial phenotypic descriptors," Genetic and Genomic Medicine, preprint, Jan. 2021. doi: 10.1101/2020.12.28.20248193.

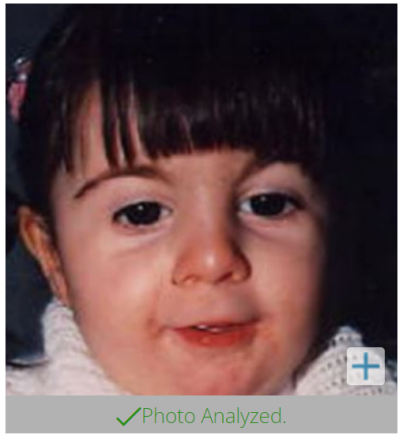
FACE2GENE CLINIC FORUMS LIBRARY RESEARCH

CLINIC | CASE LIST | NEW CASE | SUPPORT | Prof. Hilde Peeters

Add Name Or ID CASE 614698

Exam Visit | Overview | Share

Files & Photos



Suggested Phenotypic Features

- Global developmental delay...
- Short stature
- Thin upper lip vermilion...
- Micrognathia
- Smooth philtrum
- Sporadic
- Low-set ears
- Epicanthus
- Microcephaly
- Autosomal dominant inherit...

Analyze Clir

Add Mea

- Cornelia De Lange Syndrome...
- Rubinstein-Taybi Syndrome...
- Coffin-Siris Syndrome...
- Chromosome 8, Distal Del 8p...
- Auriculocondylar Syndrome...
- Mandibulofacial Dysostosis, Gu...
- Arthrogryposis, Distal, Type 2...
- Mosaic Trisomy 16
- Macroblepharon - Macrostomia...
- Pierre Robin Sequence with Fac...
- Chromosome 17q21.31 Duplicatio...

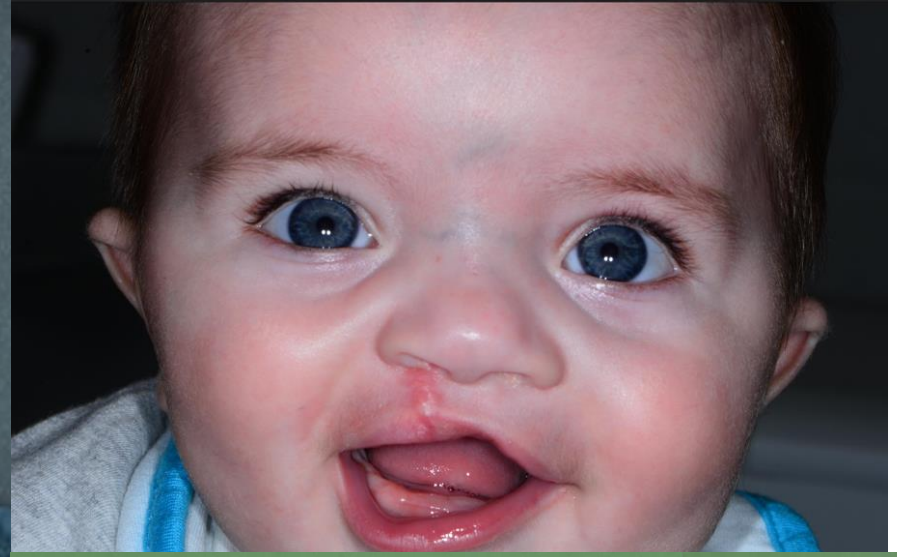
Case Analysis

PROCESS GRAPHICAL VIEW

Rare | Ultra-Rare BETA | Undiagnosed BETA

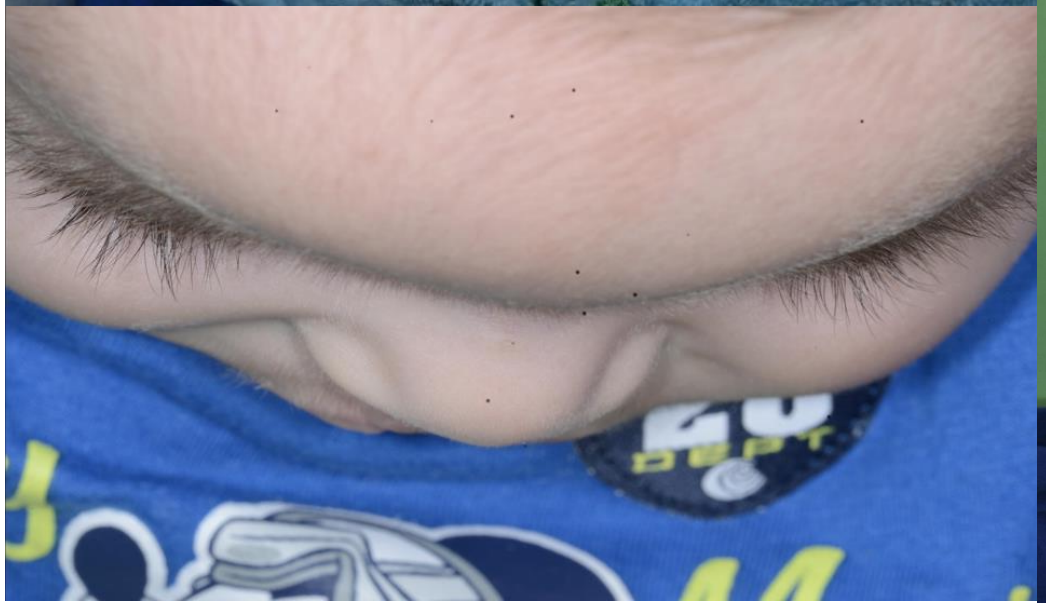
Show facial analysis only

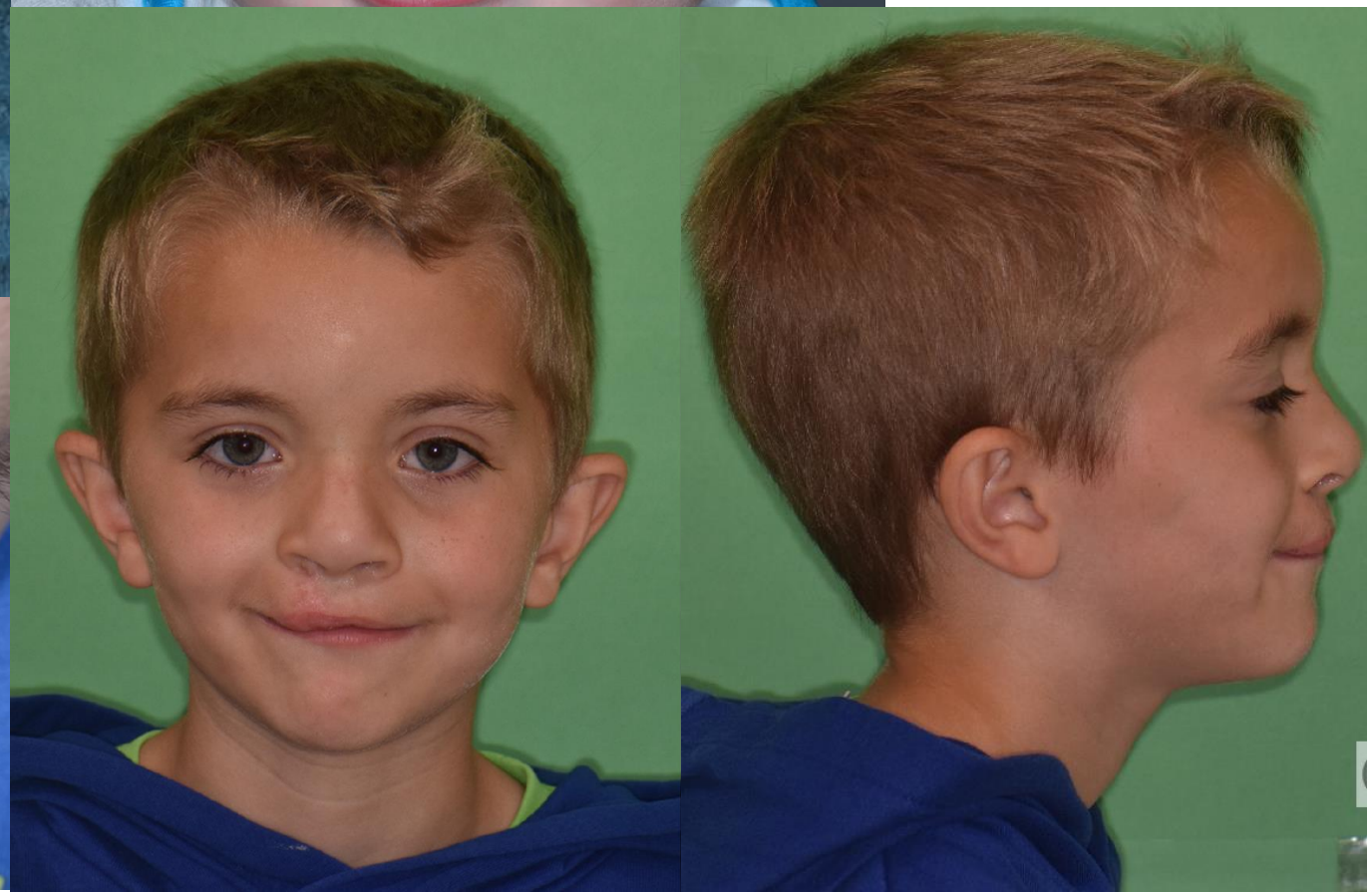
Add Syndromes >

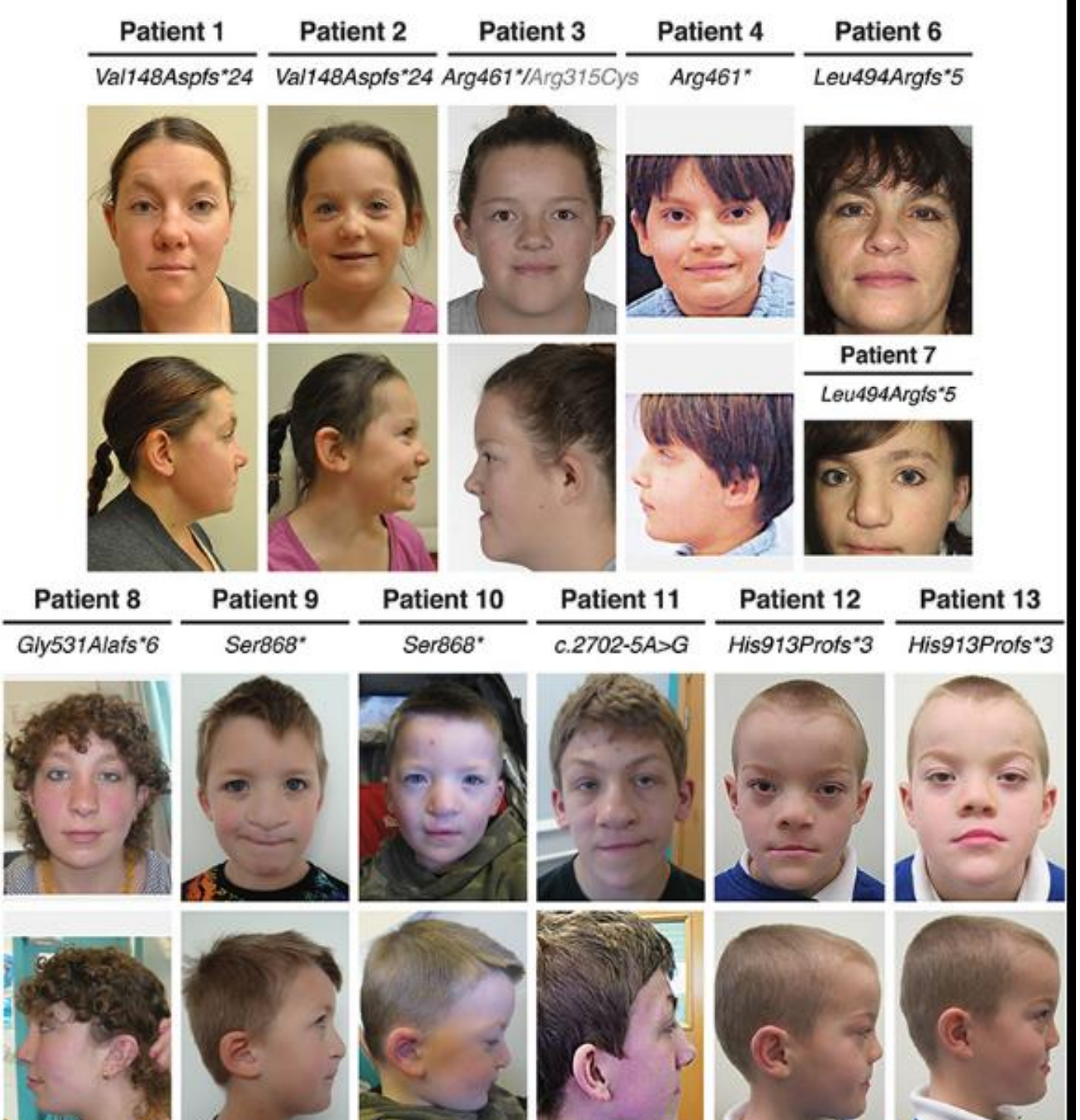


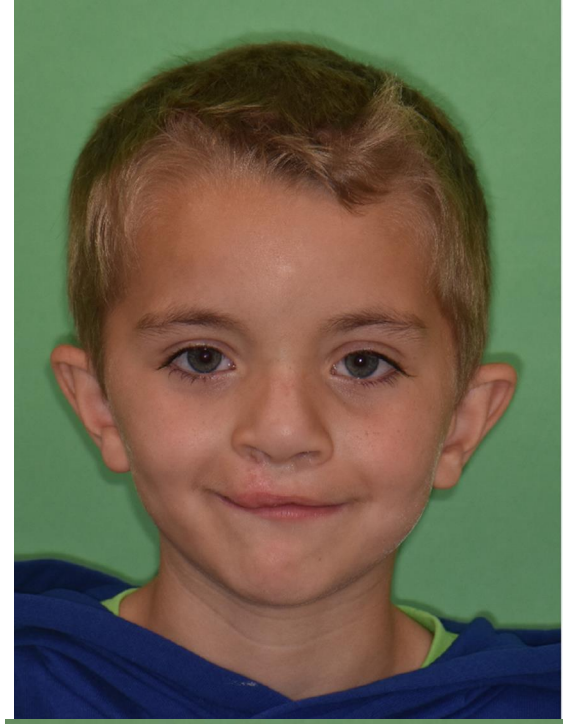
De novo c.1180T>C (p.Cys394Arg) in CTNND1

blepharocheilodontic (BCD) syndrome









Hum Mol Genet. 2020 Jul 21;29(11):1900-1921.

CASE LIST

HIPAA & GDPR COMPLIANT

Add name or ID CASE 1054239

Exam Visit | Overview | Share with Lab

Files & Photos

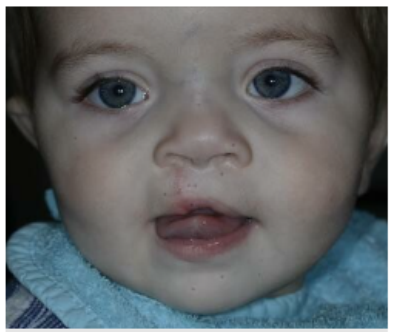


Photo Analyzed.

Suggested Phenotypic Features

- Long philtrum
- Neurogenic bladder
- Fetal megacystis
- Urinary incontinence
- Urinary hesitancy
- Nocturia
- Urinary retention
- Bladder diverticulum
- Abnormality of the bladder...
- Hypoplasia of the uterus...

REFINE

Analyze Clinical Note Beta >

Add Measurements >

View Facial D-Score Beta >

Case Analysis

Search Gene

Graphical View >

For better analysis, please review all ranked syndromes, not only the top ones Don't show this again

Rare | Ultra-Rare Beta | Undiagnosed Beta

Top Genes: FGFR3 FGFR2 TWIST1 H19 MLH1 ELN ERCC4 GABRB3 IGF2 GL

Add Syndromes >

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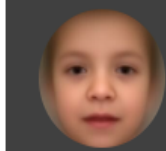
Rare | Ultra-Rare Beta | Undiagnosed Beta

Top Genes: FGFR3 FGFR2 TWIST1 H19 MLH1 ELN ERCC4 GABRB3 IGF2 GL

Add Syndromes >

Noonan Syndrome; NS



HIGH MED LOW

Differential
Clinically Diagnosed
Molecularly Diagnosed

GESTALT FEATURE

Congenital Contractures Of The Limb...A, And Developmental Delay; CLIFAHDD

HIGH MED LOW

Differential
Clinically Diagnosed
Molecularly Diagnosed

GESTALT FEATURE

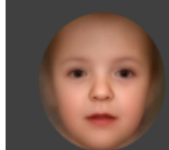
Cornelia De Lange Syndrome

HIGH MED LOW

Differential
Clinically Diagnosed
Molecularly Diagnosed

GESTALT FEATURE

Muenke Syndrome; MNKES

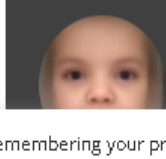


HIGH MED LOW

Differential
Clinically Diagnosed
Molecularly Diagnosed

GESTALT FEATURE

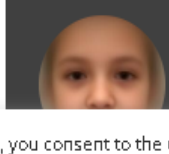
Robinow Syndrome, Autosomal Dominant



HIGH MED

Differential
Clinically Diagnosed

Neurofibromatosis, Type I; NF1



HIGH MED

Differential
Clinically Diagnosed

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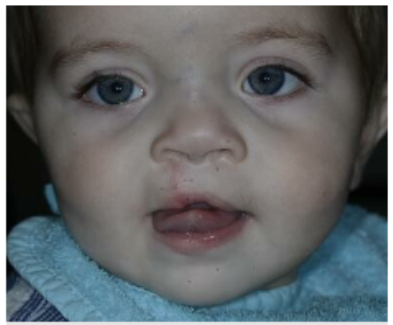
HIPAA & GDPR COMPLIANT

Add name or ID

CASE 1054239

Exam Visit | Overview | Share with Lab

Files & Photos



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- Bladder diverticulum
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- Hypoplasia of the uterus...

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Analyze Clinical Note Beta >

Add Measurements >

View Facial D-Score Beta >

Case Analysis

Search Gene

Graphical View >

Rare | Ultra-Rare Beta | Undiagnosed Beta

Top Genes: SLC3A1 KRAS SMO PPM1B CCBE1 PAM16

Spondylometaphyseal Dysplasia, Megarbane-Dagher-Melike Type; SMDMDM

HIGH MED LOW GESTALT SIMILARITY

2 Patients

Differential

Clinically Diagnosed

Molecularly Diagnosed

Intellectual Disability - Nalcn Mutations

HIGH MED LOW GESTALT SIMILARITY

1 Patient

Differential

Clinically Diagnosed

Molecularly Diagnosed

Cleft Lip/Palate With Characteristi...And Lethal Congenital Heart Disease

HIGH MED LOW GESTALT SIMILARITY

2 Patients

Differential

Clinically Diagnosed

Molecularly Diagnosed

Curry-Jones Syndrome; CRJS

HIGH MED LOW GESTALT SIMILARITY

1 Patient

Differential

Clinically Diagnosed

Molecularly Diagnosed

Chromosome 5p15 Microdeletion

HIGH MED LOW GESTALT SIMILARITY

2 Patients

Differential

Clinically Diagnosed

Molecularly Diagnosed

Chudley-Mccullough Syndrome

HIGH MED LOW GESTALT SIMILARITY

2 Patients

Differential

Clinically Diagnosed

Molecularly Diagnosed

Hydrops Fetalis, Idiopathic

HIGH MED LOW

1 Patient

Differential

Clinically Diagnosed

Shprintzen Omphalocele Syndrome

HIGH MED LOW

3 Patients

Differential

Clinically Diagnosed

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Exam Visit | Overview | Share with Lab

Files & Photos

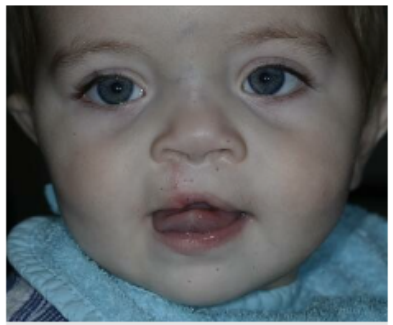


Photo Analyzed.

Suggested Phenotypic Features

- Long philtrum
- Neurogenic bladder
- Fetal megacystis
- Urinary incontinence
- Urinary hesitancy
- Nocturia
- Urinary retention
- Bladder diverticulum
- Abnormality of the bladder...
- Hypoplasia of the uterus...

REFINE

Analyze Clinical Note Beta >

Add Measurements >

View Facial D-Score Beta >

Case Analysis

Search Gene

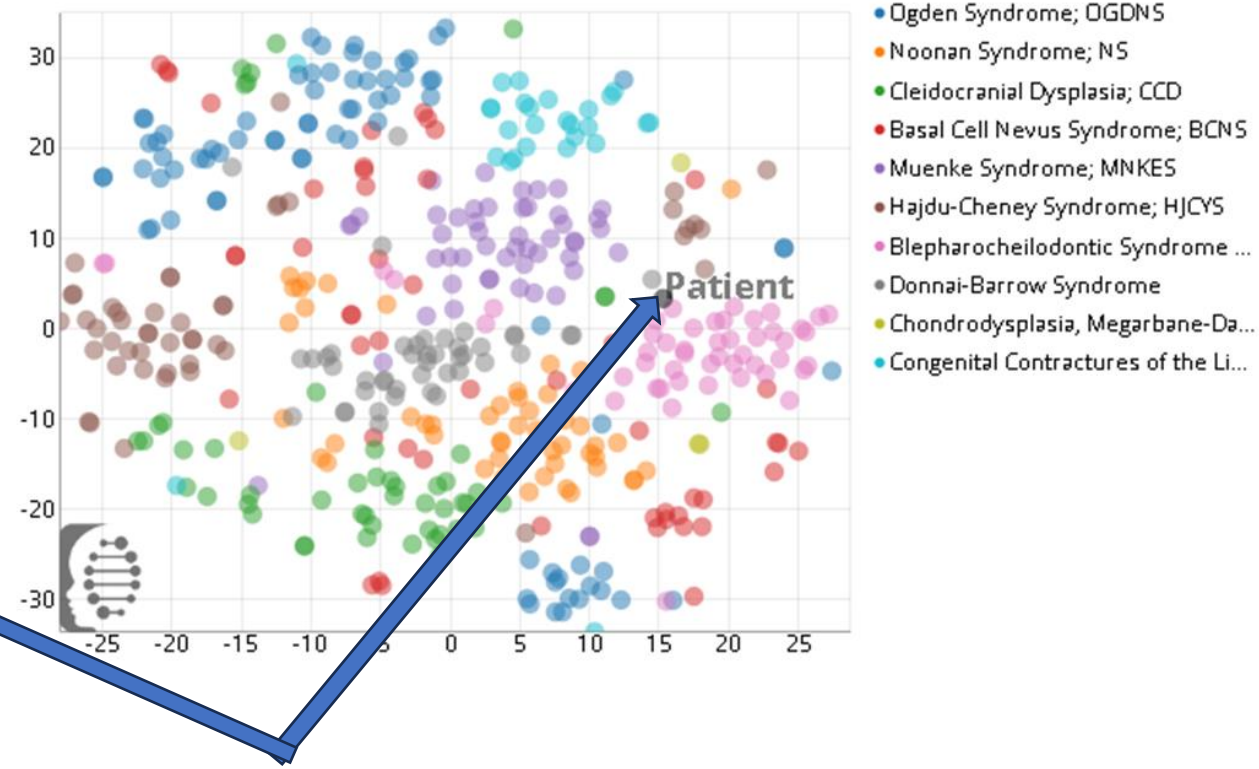
Graphical View >

Rare | Ultra-Rare Beta | Undiagnosed Beta

Top Genes: SLC3A1 KRAS SMO PPM1B CCBE1 PAM16

t-SNE Visualization: Case Photo

View in Fullscreen



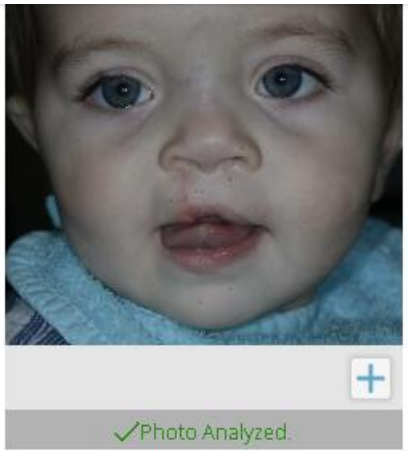


Photo Analyzed

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REFINE

Analyze Clinical Note Beta >

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For better analysis, please review all ranked syndromes, not only the top ones

Don't show this again

Rare | Ultra-Rare Beta | Undiagnosed Beta

Match my patient

SELECTED PATIENT (1)

Patient 757387

HIGH
MED
LOW
GESTALT SIMILARITY

Select Patient

Features:
Cleft palate

Contact Clinician >

Undiagnosed Matched Patient 757387

CANCEL APPLY

Case photo

Visualization

GRAPHICAL VIEW

< BACK

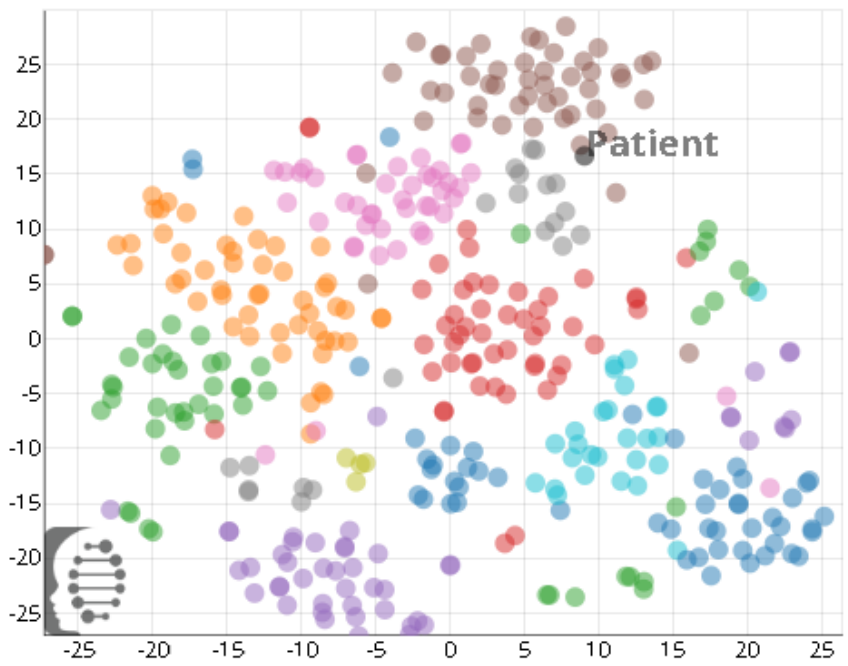
Similarity 0.9

HIGH
MED
LOW
GESTALT SIMILARITY

Select Patient

t-SNE Visualization: Matched Patient Photo

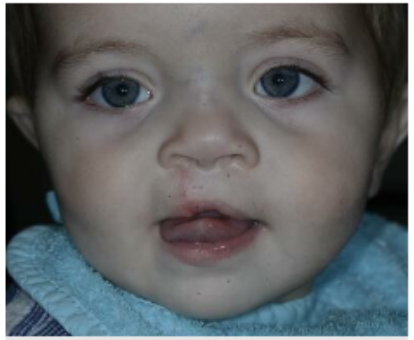
View in Fullscreen



- Ogden Syndrome; OGDNS
- Noonan Syndrome; NS
- Cleidocranial Dysplasia; CCD
- Muenke Syndrome; MNKES
- Hajdu-Cheney Syndrome; HJCYS
- Blepharocheilodontic Syndrome 1...
- Donnai-Barrow Syndrome
- Craniofacial Dysmorphism, Skelet...
- Chondrodysplasia, Megarbane-D...
- Congenital Contractures of the Li...

Exam Visit | Overview | Share with Lab

Files & Photos



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- Hypoplasia of the uterus...

REFINE

Analyze Clinical Note Beta >

Add Measurements >

View Facial D-Score Beta >

Case Analysis

CTNND1

Graphical View >

Rare | Ultra-Rare Beta | Undiagnosed Beta

Top Genes: FGFR3 FGFR2 TWIST1 H19 MLH1 ELN ERCC4 GABRB3 IGF2 G

Add Name Or ID CASE 1054239

Ogden Syndrome; OGDNS

Lissencephaly 1; LIS1

Beckwith-Wiedemann Syndrome; BWS

20

Blepharocheilodontic Syndrome 1; BCDS1

Smith-Magenis Syndrome; SMS

Megalencephaly-Capillary Malformation-Polymicrogyria Syndrome; MCAP

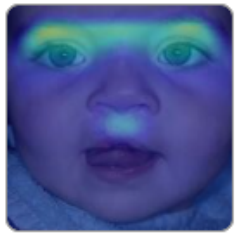
Aarskog-Scott Syndrome; AAS

Intellectual Developmental Disorder, X-Linked Syndromic, Lubs Type; MRXSL

Blepharocheilodontic Syndrome 1; BCDS1

CANCEL APPLY

Image Comparison


CASE PHOTO


COMPOSITE PHOTO

HEAT MAP

SPLIT VIEW

Similarity

HIGH
MED
LOW

GESTALT FEATURE

Diagnosis

Differential
Clinically Diagnosed
Molecularly Diagnosed

Syndrome Info (London Medical Databases)

SYNONYMS

Bcd Syndrome. Clefting. Ectropion. And Conical Teeth. Ectropion. Inferior. With Cleft Lip And/or Palate. Elschnig Syndrome. Lagophthalmia With Bilateral Cleft Lip And Palate

OMIM #

119580

LOCATION

16q22.1, 11q21.1

INHERITANCE MODE

Autosomal Dominant

A family was described in which a quarter of the putative gene carriers had a cleft lip and/or palate. 40% had ectropion of the lower lid and nearly 50% had conical teeth. In 28% only one feature was present. The young proband had sparse hair but this was not a feature in others. Rapp-Hodgkin syndrome was considered as part of the differential diagnosis. This condition may be the same as cleft lip/palate-

Typical Features | Related Genes

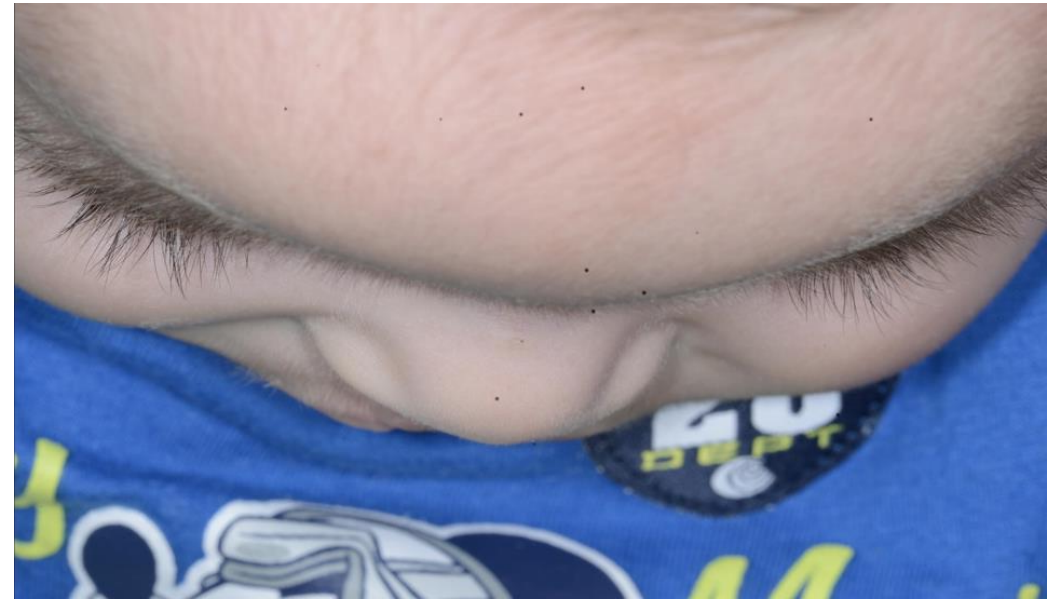
Filter Features

SELECTED (0)

TYPICALLY PRESENT (17)

- Sparse hair
- Flat face
- Ectropion of lower eyelids
- Hypodontia
- Clinodactyly
- Distichiasis
- Autosomal dominant inheritance
- Hypertelorism
- Neural tube defect
- Conical tooth
- Small nail
- High forehead
- Cleft upper lip
- High anterior hairline
- Choanal atresia
- Anal atresia

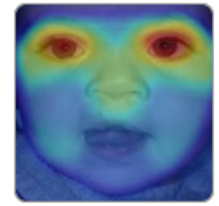
TYPICALLY ABSENT (0)



Noonan Syndrome; NS

CANCEL APPLY

Image Comparison


CASE PHOTO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Molecularly Diagnosed

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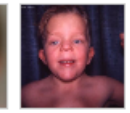


SYNONYMS

Typical Features | Related Genes

Filter Features

SELECTED (0)

TYPICALLY PRESENT (282)

- Asymmetry of the thorax

Laboratory of
IMAGING GENETICS

The Leuven 3D Facial Dysmorphology Project

Applications of 2D and 3D imaging

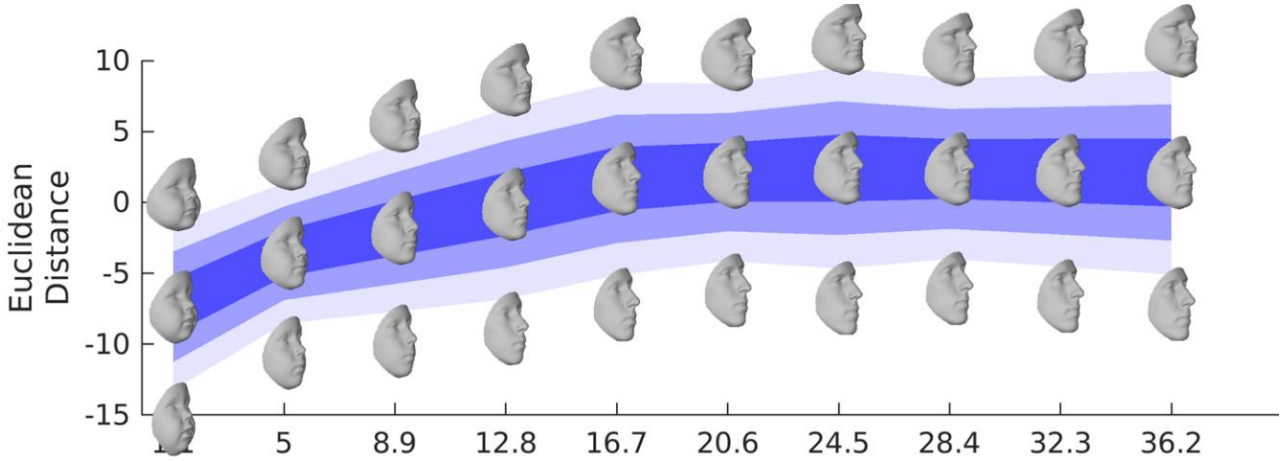
- Syndrome recognition, classification, ranking
- Explore the phenotypic variability of syndromic conditions
    - Monogenic conditions: study the relation of the facial phenotype with molecular diagnosis
    - Expand on the phenotypic spectrum -> milder phenotype
    - Variant interpretation

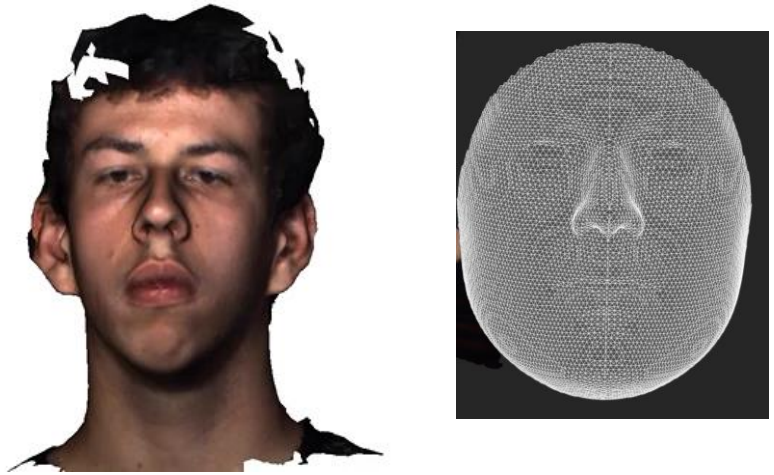
How to assess an individual face?

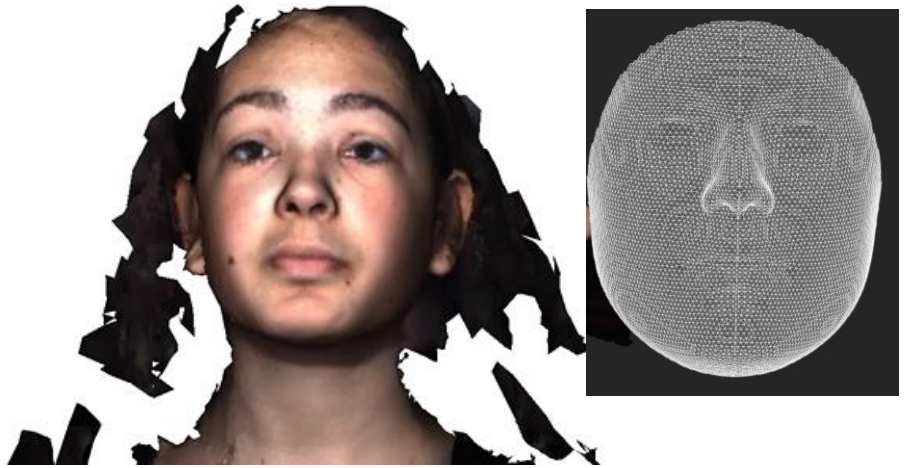
Article | Open Access | Published: 09 June 2021

Large-scale open-source three-dimensional growth curves for clinical facial assessment and objective description of facial dysmorphism

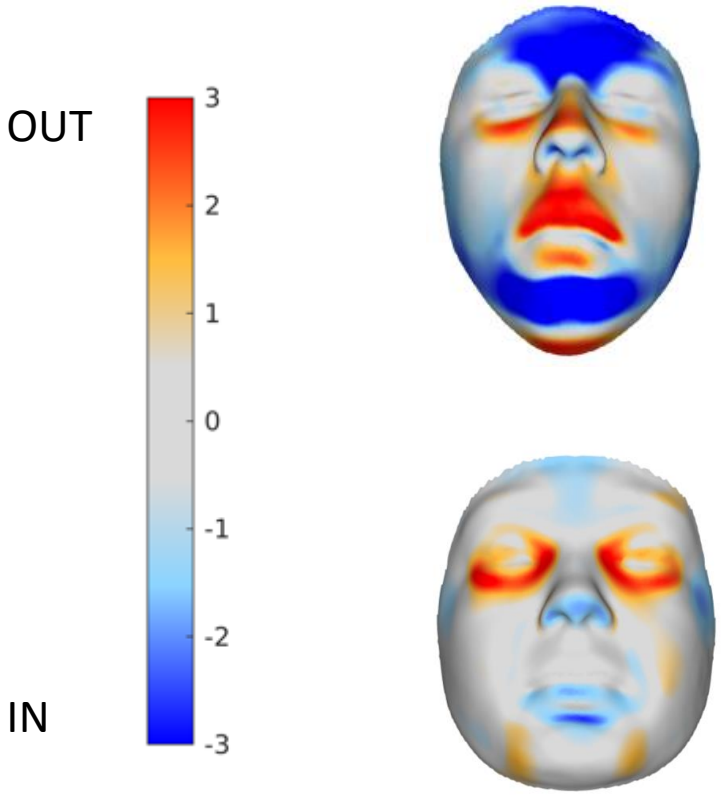
Harold S. Matthews, Richard L. Palmer, Gareth S. Baynam, Oliver W. Quarrell, Ophir D. Klein, Rich Spritz, Raoul C. Hennekam, Susan Walsh, Mark Shriver, Seth M. Weinberg, Benedikt Hallgrimsson, P Hammond, Anthony J. Penington, Hilde Peeters & Peter D. Claes

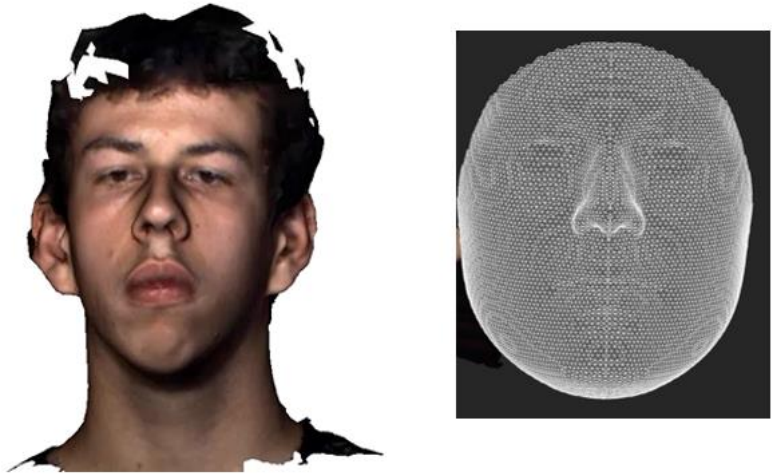


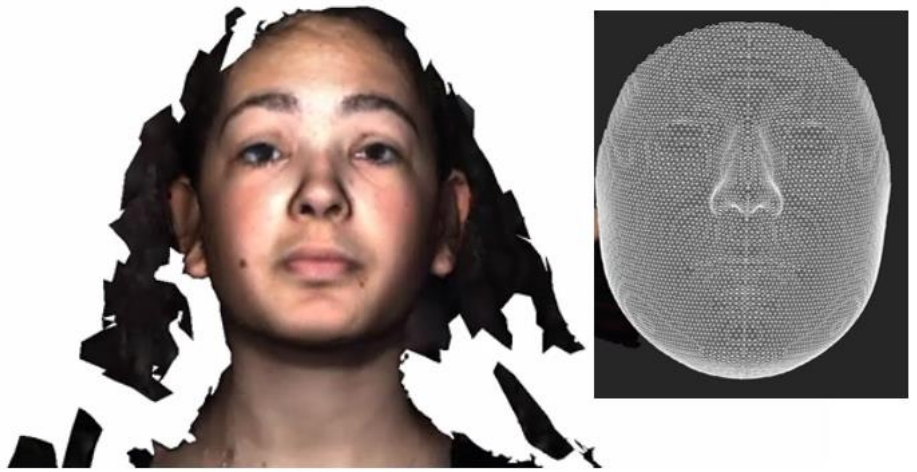




How to assess an individual face?





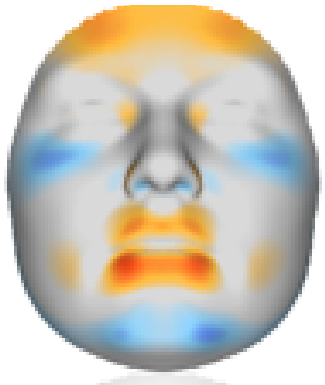


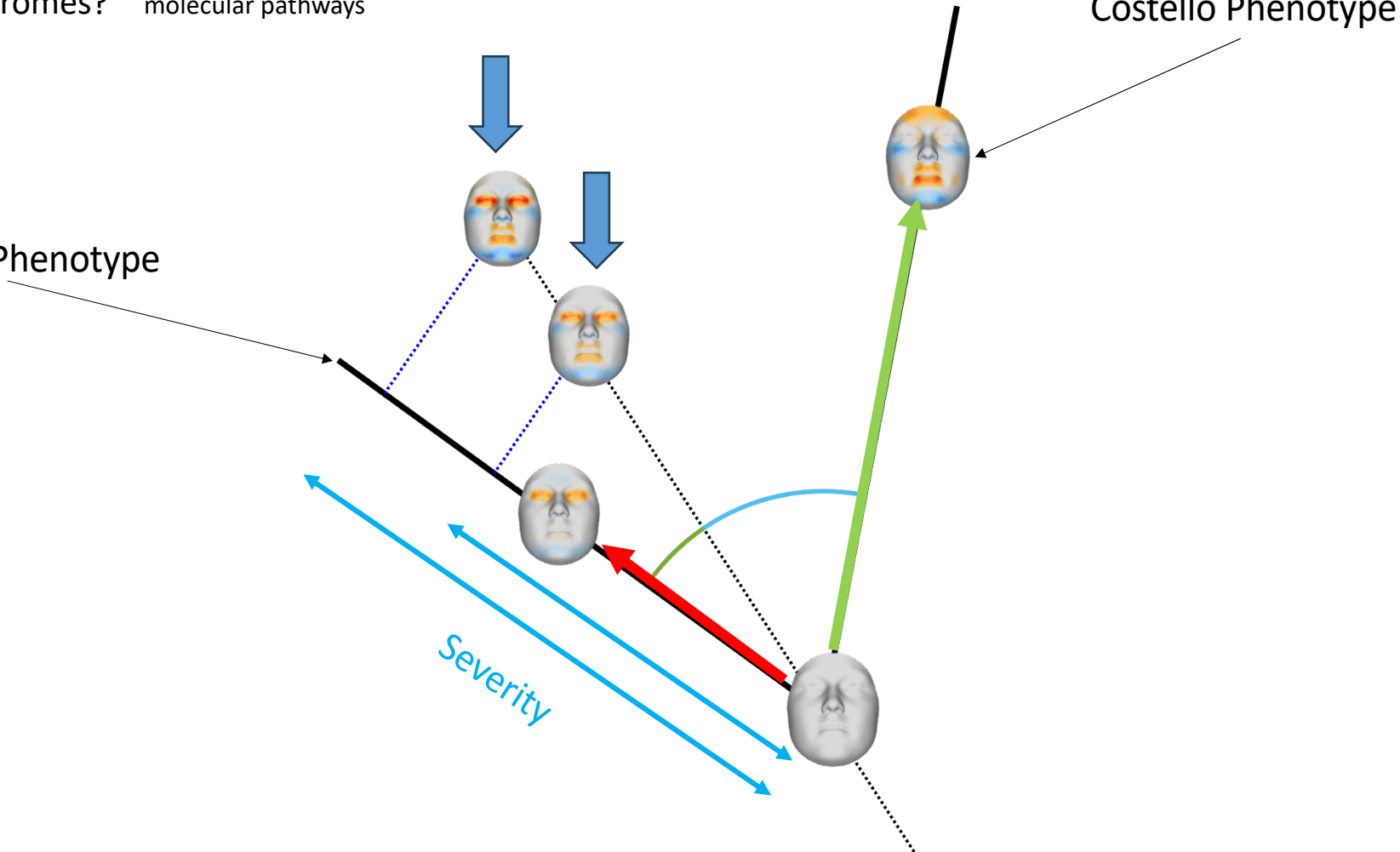
Genotype phenotype correlation: syndrome delineation, discrimination, individual patient evaluation

- What parts of the face are affected and in what ways?
- Can we define a degree of severity for individual patients?
- Can we measure similarity between syndromes? ~ molecular pathways

Average Noonan signature

Average Costello signature





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J Med Genet. 2022 Jul 20;jmedgenet-2021-108366. doi: 10.1136/jmedgenet-2021-108366.
Online ahead of print.

Refining nosology by modelling variation among facial phenotypes: the RASopathies

Harold Matthews [1 2 3], Michiel Vanneste [1 2], Kaitlin Katsura [4], David Aponte [5], Michael Patton [6], Peter Hammond [1], Gareth Baynam [7 8 9 10], Richard Spritz [11], Ophir D Klein [4], Benedikt Hallgrimsson [5], Hilde Peeters [1], Peter Claes [12 2 3 13]

Genotype phenotype correlation: syndrome delineation, discrimination, individual patient evaluation

- What parts of the face are affected and in what ways?
- Can we define a degree of severity for individual patients?
- Can we measure similarity between syndromes? ~ molecular pathways

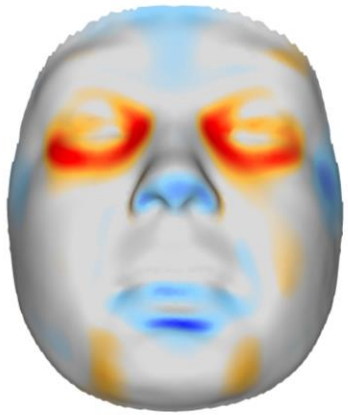
New syndromes ?

aspecific facial phenotype

or

to rare to become recognizable by either of us

What is it in the face that gives away the major gene effect?



~ Mendelian cause?

Applications of 2D and 3D imaging

- Syndrome recognition, classification, ranking

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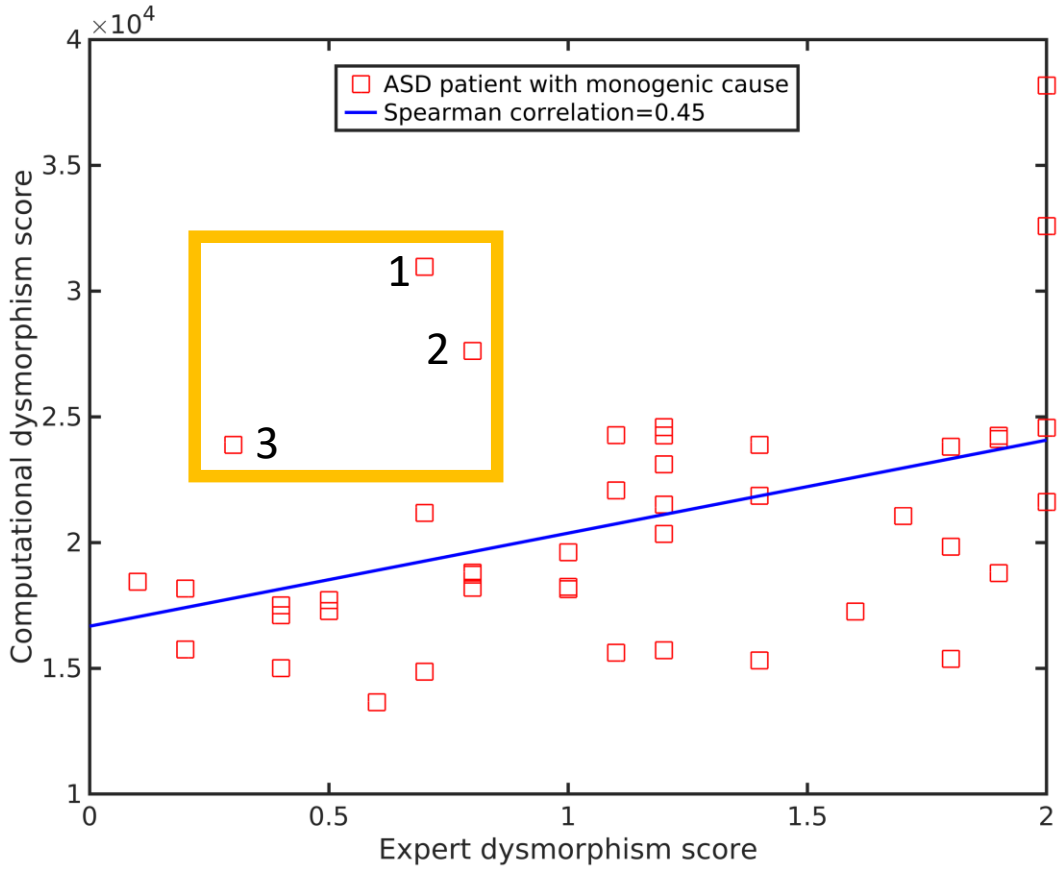
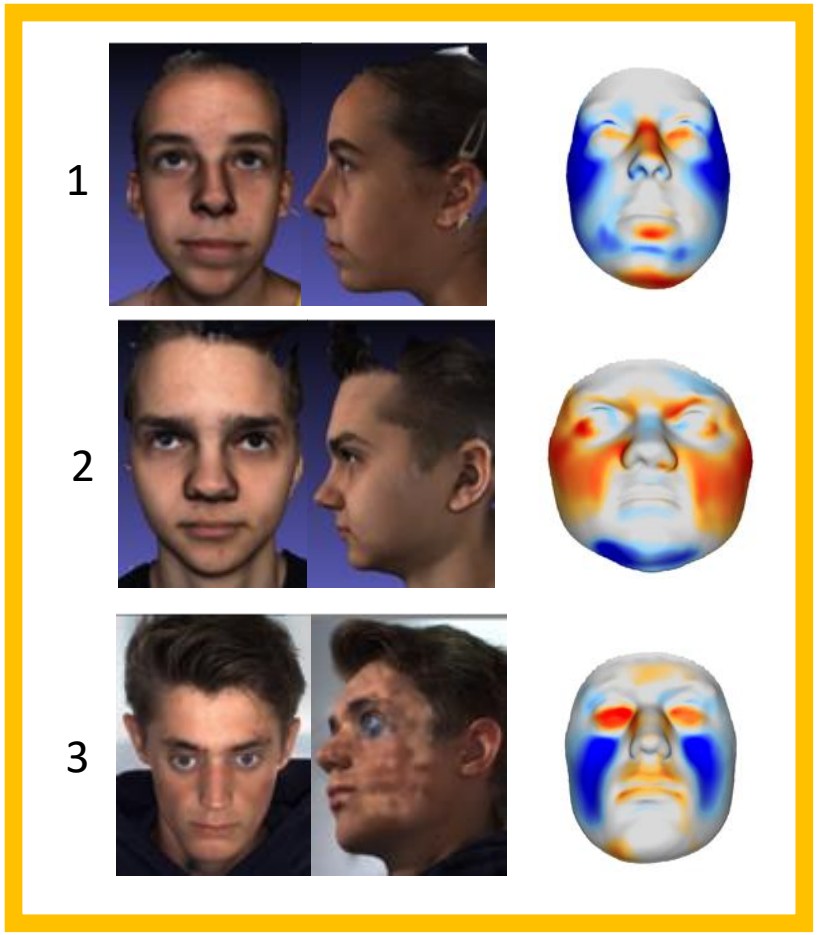
    - Monogenic conditions: study the relation of the facial phenotype with molecular diagnosis
    - Expand on the phenotypic spectrum -> milder phenotype
    - Variant interpretation

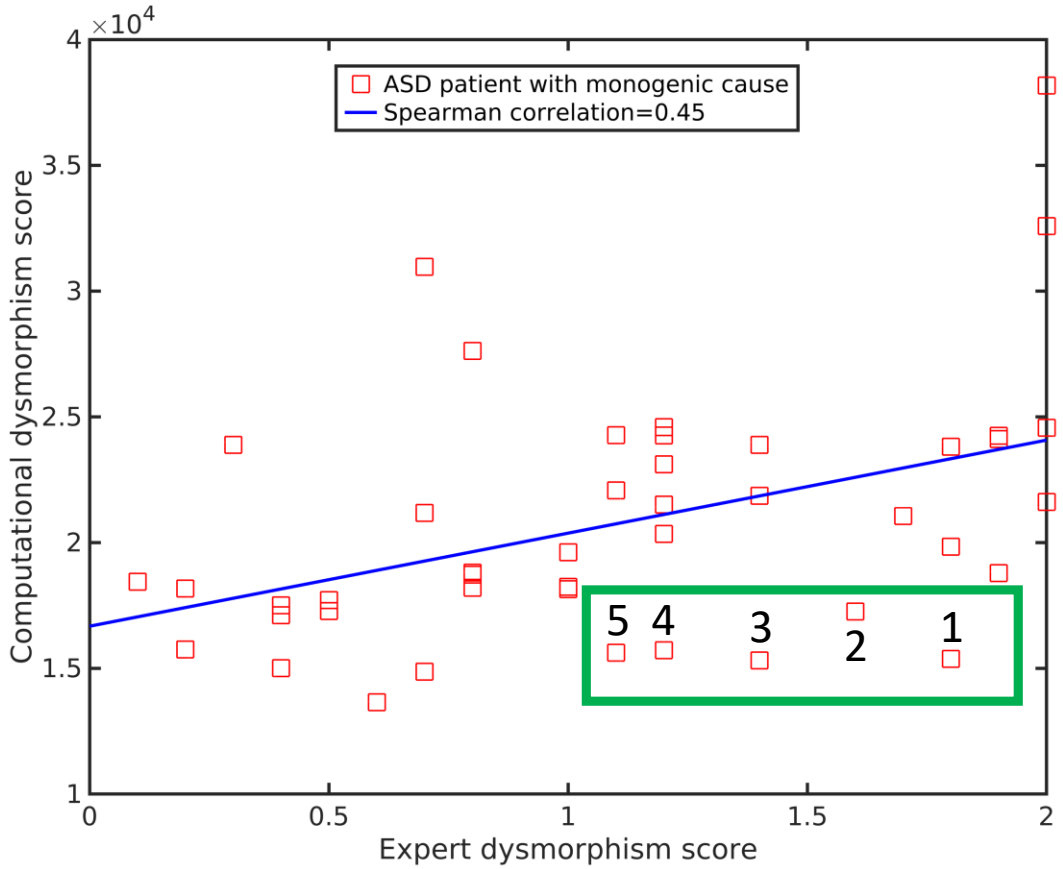
- Normal versus abnormal facial variation

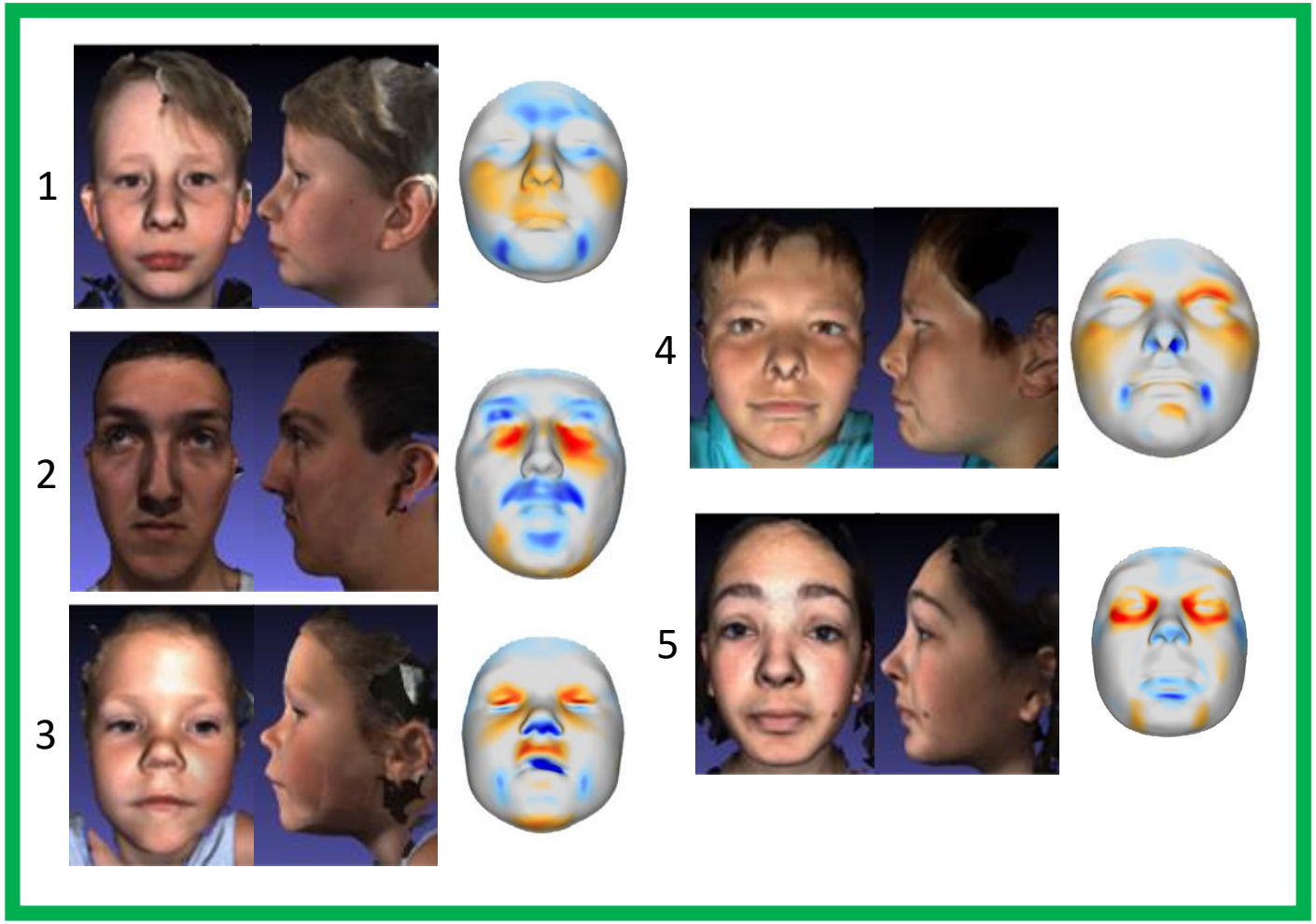
Computational versus expert dysmorphism scores in ASD patients with monogenic causes





Computational versus expert dysmorphism scores in ASD patients with monogenic causes





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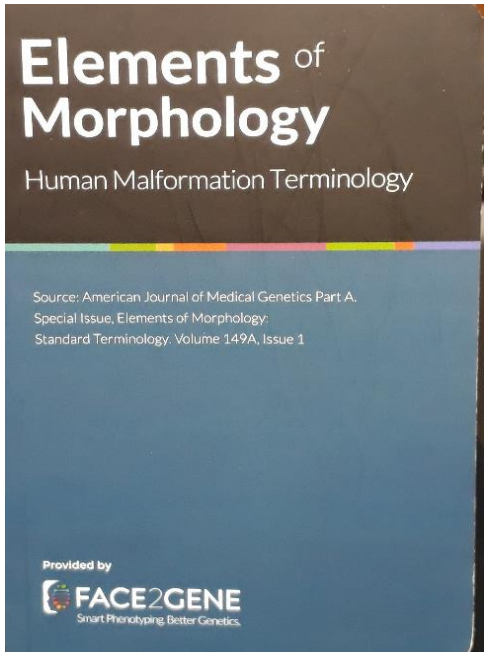
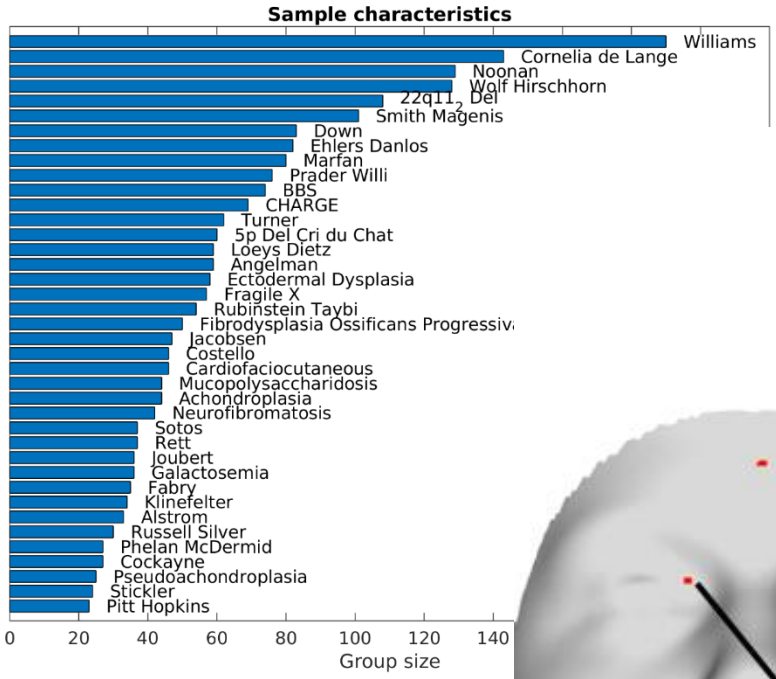
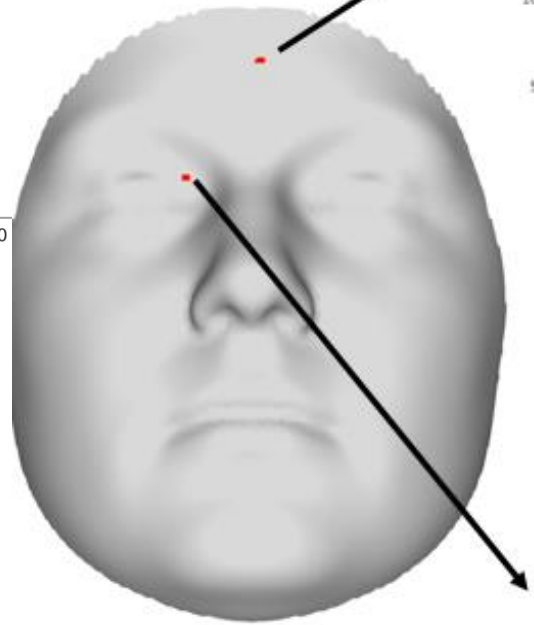
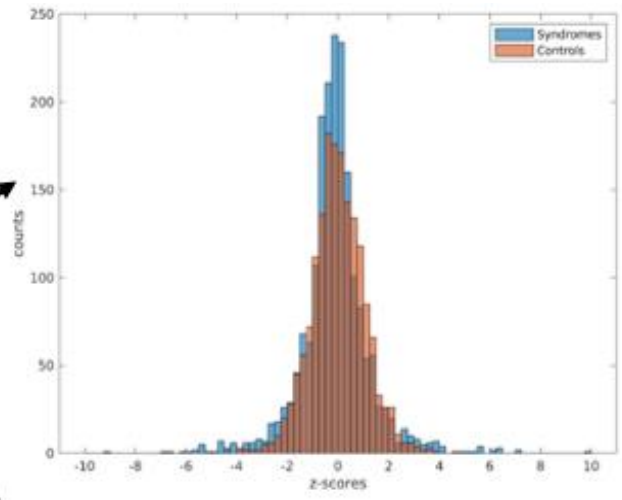
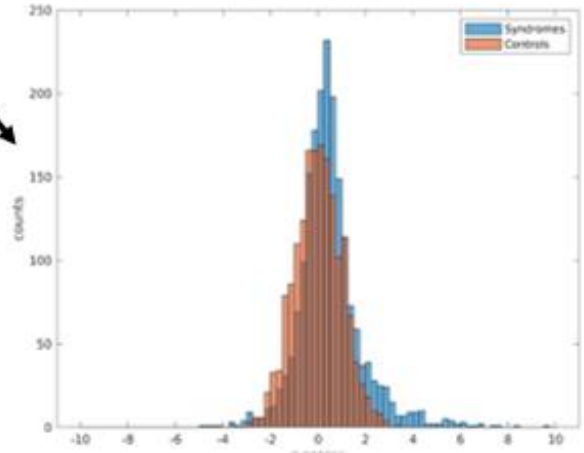


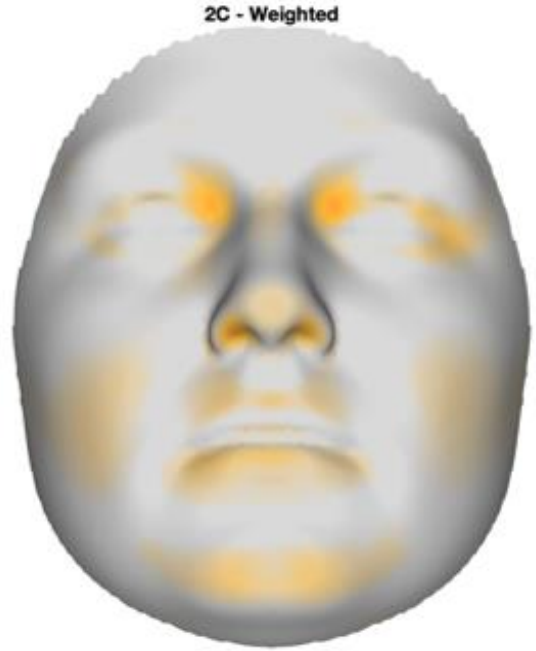

Sample characteristics
Williams
Cornelia de Lange
Noonan
Wolf Hirschhorn
22q11.2 Del
Smith Magenis
Down
Ehlers Danlos
Marfan
Prader Willi
BBS
CHARGE
Turner
5p Del Cri du Chat
Loeys Dietz
Angelman
Ectodermal Dysplasia
Fragile X
Rubinstein Taybi
Fibrodysplasia Ossificans Progressiv
Jacobsen
Costello
Cardiofaciocutaneous
Mucopolysaccharidosis
Achondroplasia
Neurofibromatosis
Sotos
Rett
Joubert
Galactosemia
Fabry
Klinefelter
Alstrom
Russell Silver
Phelan McDermid
Cockayne
Pseudoachondroplasia
Stickler
Pitt Hopkins
0 20 40 60 80 100 120 140
Group size



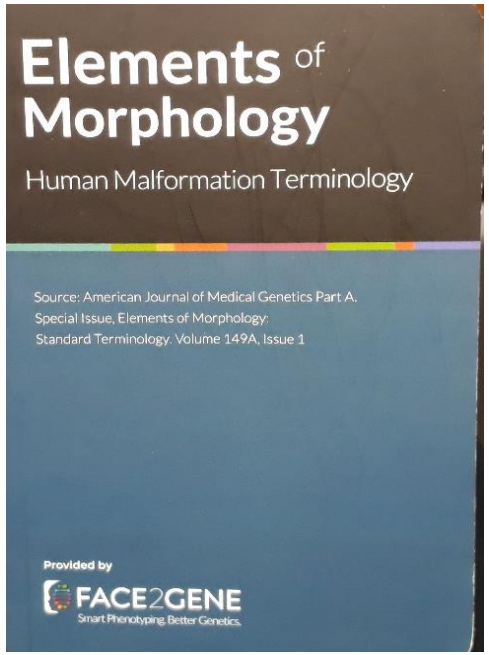

Syndromes
Controls
counts
z-scores

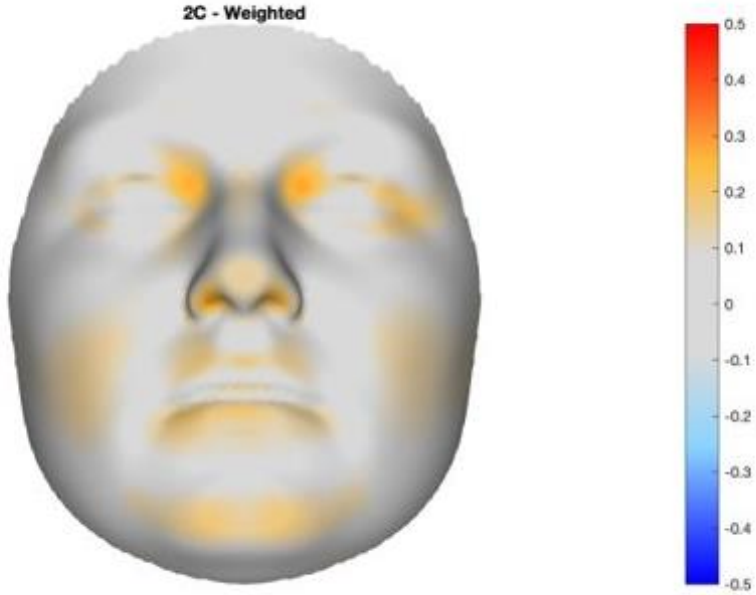
(...)


Syndromes
Controls
counts


2C - Weighted

Computational versus expert dysmorphism scores in ASD patients with monogenic causes





Files & Photos

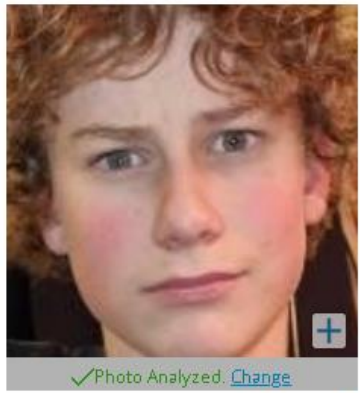


Photo Analyzed. Change

2 OF 2

Suggested Phenotypic Features

- Deeply set eye
- Striae distensae
- Pectus carinatum
- Pectus excavatum
- Dental crowding
- Myopia
- Retinal detachment
- Cataract
- Glaucoma
- Downslanted palpebral fiss...

REFINE

Analyze Clinical Note Beta >

View Facial D-Score Beta >

CLINIC FORUMS LIBRARY RESEARCH

SUPPORT Prof. Hilde Peeters

2Gene Pediatrician view

Examples

Later bekijken Delen

Karen Gripp

Case Analysis

Search Gene

Graphical View >

Rare

Ultra-Rare Beta

Undiagnosed Beta

Facial D-Score Beta

LOW UNKNOWN-SIGNIFICANCE HIGH

Photo Analyzed.

Photo Analyzed.

LOW UNKNOWN SIGNIFICANCE HIGH

Medical student

LOW UNKNOWN SIGNIFICANCE HIGH

Software designer

LOW UNKNOWN SIGNIFICANCE HIGH

Smith-Lemli-Opitz syndrome

LOW UNKNOWN SIGNIFICANCE HIGH

Medical student

Applications of 2D and 3D imaging

- Syndrome recognition, classification, ranking

- Explore the phenotypic variability of syndromic conditions
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