

**5<sup>th</sup> Congress of the European Academy of Neurology**  
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**Teaching Course 10**

**Clinical science in muscle disorders (Level 2)**

## **The “myopathic” face**

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# *The “myopathic” face: a Gestalt approach*

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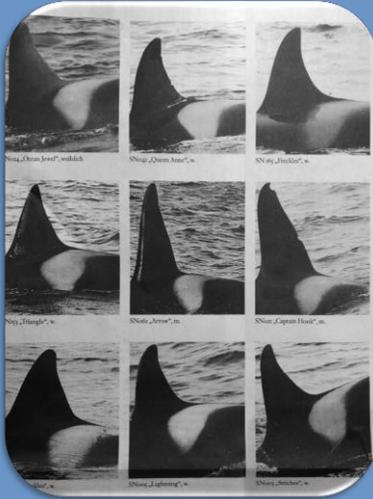


## Disclosures

- BS serves as member of the Global Advisory Boards from Amicus Therapeutics, Audentes Therapeutics, Nexien BioPharma, Lupin Therapeutics
- BS is a member of the European Pompe Registry Advisory Boards sponsored by Genzyme Europe B.V.
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- BS has received unrestricted research support from Genzyme Europe B.V. and Greenovation Biopharm
- Unreferenced data or images and movies used within this presentation are the speaker’s own.



## Gestalt = shape = phenotype



Frankfurter Allgemeine Sonntagszeitung 03.03.2019



<https://lehrerfortbildung-bw.de/flug/>



## Jizz - Gestalt

Among birdwatchers and naturalists:  
JIZZ means the characteristic impression  
given by a particular species of animal  
or plant.



Gestalt (German): describes a specific shape and design of an object



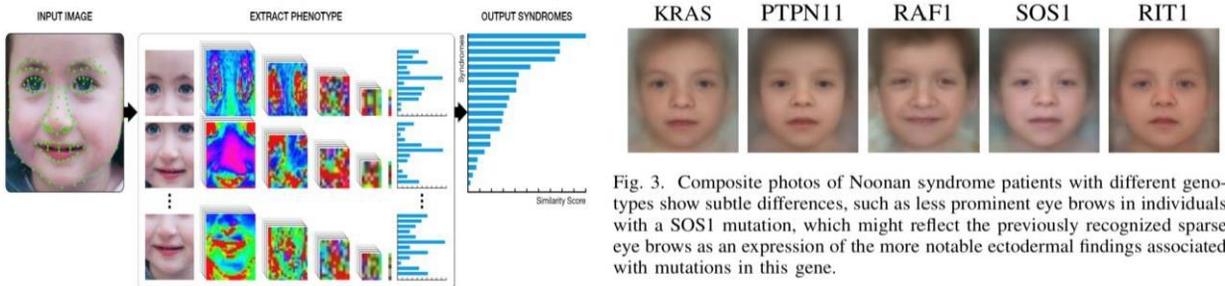
# deepGestalt

Letter | Published: 07 January 2019

## Identifying facial phenotypes of genetic disorders using deep learning

Yaron Gurovich<sup>1</sup>, Yair Hanani, Omri Bar, Guy Nadav, Nicole Fleischer, Dekel Gelbman, Lina Basel-Salmon, Peter M. Krawitz, Susanne B. Kamphausen, Martin Zenker, Lynne M. Bird & Karen W. Gripp

*Nature Medicine* 25, 60–64 (2019) | [Download Citation](#)



## The face in NMDs

- Dysmorphic pattern
- Temporal muscle atrophy
- Ptosis
- External and internal oculomotor paresis
- Buccal weakness
- Palate and Teeth deformities
- Tongue alterations



## The face in NMDs

- Dymorphic pattern **Clinical Gestalt**

- generalized myotonic myopathy
- masklike face
- blepharophimosis
- small mouth
- microretognathia
- dental malalignments
- cleft palate
- skeletal dysplasia
- contracture of joints
- growth retardation
- bone maturation delay
- stiff muscle hypertrophy
- percussion myotonia
- **Mutations in perlecan gene (HSPG2)**



Giovanni  
<https://www.youtube.com/watch?v=Lg1j3fCnZdM>

### Schwartz-Jampel Syndrome



<https://lockerdomo.com/tlc/8628403607879188>



Adv Biomed Res 2015, 4:163



## The face in NMDs

- Temporal muscle atrophy



### Myotonic dystrophy type 1 **CTG repeat expansion in the DMPK gene (rare in DM2)**



# The face in NMDs



## • Ptosis I



Posey & Spiller 1904



Dynamic or fixed ptosis?

Unilateral

- cranial nerve palsy, diabetes, tumor, etc.

Bilateral

- Borreliosis, sarcoidosis, thyroid disease, etc.
  - Myasthenia gravis (asymmetric)
  - LEMS
  - Mitochondrial myopathies (asymmetric)
  - OPMD (asymmetric)
  - Ocular myositis
- DX CNS disorders, PSP etc.



# The face in NMDs

## • Ptosis II

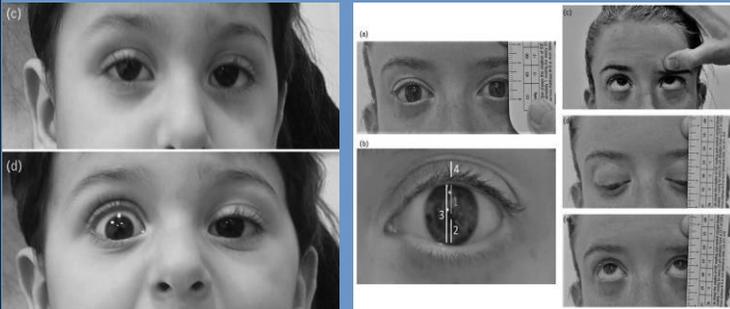


**Oculopharyngeal muscular dystrophy OPMD**, autosomal dominant GCG repeat expansion in PABPN1 gene



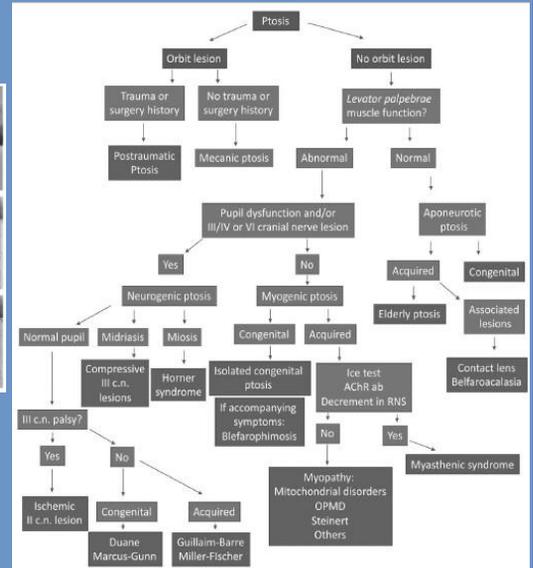
# The face in NMDs

## • Ptosis III



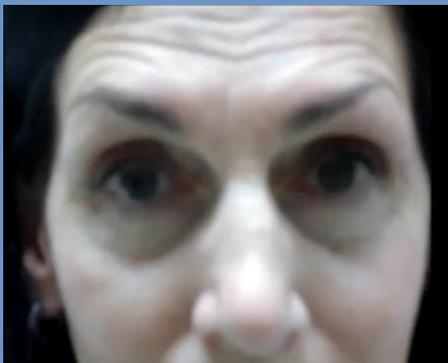
**Marcus Gunn ptosis** produced by the anomalous connection of motor fibers from the fifth to the third cranial nerve. Upper eyelid retraction upon stimulation of the pterygoid muscles, elicited by chewing

Diaz-Manera J, Lunac S, Roiga C  
Curr Opin Neurol 2018, 31:618–627



# The face in NMDs

## • External and internal oculomotor paresis I



Courtesy Wolfgang Löscher

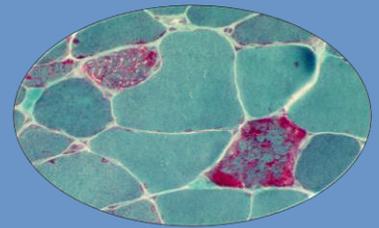


**MUSK antibody positive myasthenia gravis**

**SCN4A congenital myotonia**



## The face in NMDs



- External and internal oculomotor paresis II



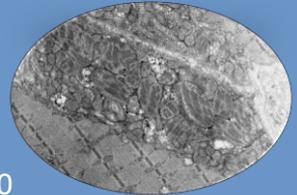
### CPEO

external ophthalmoplegia, ptosis  
facultative: myopathy



### CPEO plus

plus: non-muscular symptoms



### Kearns-Sayre syndrome

CPEO + retinitis pigmentosa, age <20  
plus: ataxia, conduction block, increased CSF protein content



## The face in NMDS

- Buccal weakness



CMD



FSDH2



FSDH1





# The face in NMDs

## Palatete, teeth and tongue alterations



- Gothic high-arched palate?
- Swallowing difficulties?
- Macroglossia?
- Tongue myotonia?
- Tongue atrophy?
- Fasciculations?
- Teeth deformities?



# The face in NMDs

## Palate and Teeth deformities

**Nemaline myopathies, Central cores disease,**

**Multiminicore disease**

Mutations in RYR1, ACTA1, TPMB, TPM2, etc.



**Congenital fiber-type disproportion**

**Crowded teeth**



**Central core myopathy**



**Gothic high-arched palate**



Elements of Morphology  
National Human Genome Research Institute



## The face in NMDs

### Tongue alterations

- **Tongue hypertrophy**

DMD, Pompe, Amyloid, LGMD2I, BSMA early



Suggestive but non-specific of a dystrophic process



**Congenital myotonia**  
Mutation in the SCN4A gene



**Bulbospinal muscular atrophy Type Kennedy**  
expansion of CAG repeat in the androgen receptor gene



## Summary

- Describe what you see!
- Have a second look at different time points, e.g. of the day
- “Exercise” your patient
- Taking a family history remains helpful
- Start with currently treatable disorders (“treataboloome”)



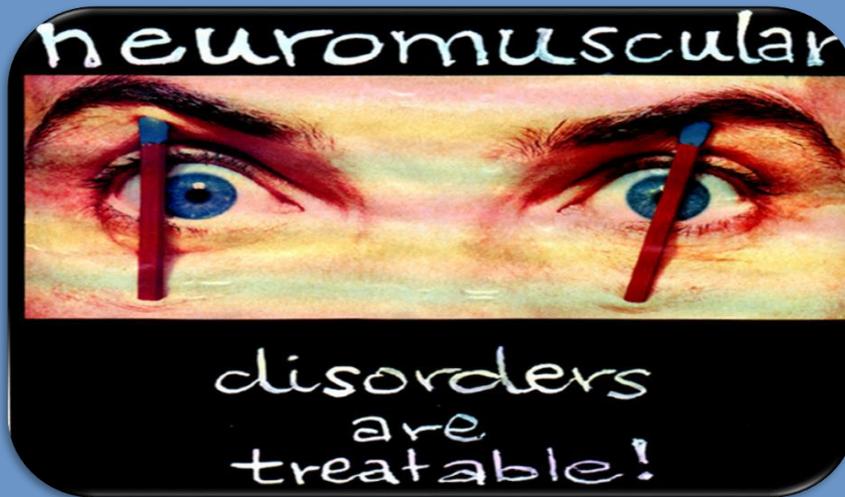
A final Quiz:  
What is the name of this clinical sign?

**Beevor sign!**

Charles Edward Beevor 1854-1908



Thank you for your attention!



[www.baur-institut.de](http://www.baur-institut.de)