Translation from Romanian

Marcus-Gunn Syndrome

DEFINITION

Marcus-Gunn syndrome is characterized by ***ptosis*** associated with ***maxillo palpebral synkinesis.***

Synonyms: ***Jaw-winking syndrome; Mandibular-eyelid synkinesis syndrome - ptosis; the Marcus-Gunn phenomenon;***

PREVALENCE

- ✓ The syndrome is responsible for **5%** of congenital ptosis.
- ✓ The etiology is still uncertain; it occurs sporadically, usually **unilaterally,** but bilateral cases have also been reported.
- ✓ It affects women and men equally and there is no racial predisposition.

CLINICAL ASPECTS

→ Ptosis is corrected by opening the mouth, lateral movements of the lower jaw or chewing, sucking, swallowing, protrusion of the tongue. This may be due to the aberrant innervation of the lifting muscle of the upper eyelid, by a branch of the mandibular division of the trigeminal nerve. This aberrant innervation has been demonstrated by **electromyographic studies** that have shown simultaneous contraction of the external pterygoid muscle and the lifting muscle of the upper eyelid.

→ **Marcus Gunn syndrome** is usually seen from birth when parents notice the retraction movement of the baby's upper eyelid during breastfeeding. It seems that the phenomenon can improve over time because the affected people learn how to compensate for the lifting of the upper eyelid during the movement of the mandible.

→ Although it does not usually affect vision and does not pose significant risks, it should be noted that Marcus Gunn syndrome is associated with various eye conditions such as strabismus, anisometropia and / or amblyopia.

- ✓ **Strabismus**, occurs in more than **50%** of cases of Marcus Gunn syndrome. Strabismus can be caused by both paralysis of the upper right muscle and double paralysis of the upper and lower right muscles.
- ✓ **Anisometropia** is a difference in diopters (over 2.5 D) between the two eyes and occurs in a proportion of **25%** in people with Marcus Gunn syndrome.
- ✓ **Amblyopia,** also called "lazy eye" is a decrease in visual acuity without an organic cause, and occurs in a proportion of **30-60%** in people with Marcus Gunn syndrome. It is important to mention that amblyopia occurs in the first years of life, when binocular vision is formed and is reversible until the age of 6-8 years; after this period, it becomes permanent. Amblyopia is a complication of the syndrome and can usually be caused by strabismus or uncorrected anisometropia; rarely, it can be caused by severe ptosis.

→ **Marcus Gunn syndrome** may also be associated with other eye conditions such as **retinitis pigmentosa or Duane syndrome.**

→ **Marcus Gunn syndrome has** also been described in **congenital central hypoventilation syndrome.** A mutation in the **PHOX2B** gene appears to affect the normal expression of this gene in the oculomotor nucleus of the midbrain.

GENETICAL ASPECTS

→ This syndrome is generally sporadic, but cases of **autosomal dominant transmission** have also been reported.

DIAGNOSIS

→ The diagnosis is established on the basis of signs, symptoms and clinical and paraclinical examination, within an ophthalmological consultation.

→ Regarding the history of the disease, it is important to ask if the syndrome was present at birth or if the symptoms occurred following an intervention in the ocular and / or facial area:

- strabismus correction operations
- eyelid surgery
- trauma (aberrant regeneration of the trigeminal nerve)

The eye exam includes:

- ✓ determination of visual acuity: it is essential to exclude amblyopia
- ✓ examination of the anterior segment of the eyeball
- ✓ cycloplegia refractometry: to exclude anisometropia

The physical exam aims to:

- ✓ **the head position:** due to eyelid ptosis, the child may keep the head and / or chin slightly raised to compensate for vision loss, a position called **ocular torticollis;** if the ptosis is significant and the child does not keep his head up, amblyopia should be considered.
- ✓ **ocular motility** (extrinsic muscles of the eyeball)
- ✓ **cover test:** to exclude a paralysis of the upper right muscle or double paralysis of the right muscles (upper and lower)
- ✓ **Bell phenomenon** (movement of the eye up and out when trying to close the eyelids): may be decreased due to paralysis of the upper right

TREATMENT

→ When required, the treatment of Marcus Gunn syndrome is surgical and consists of eyelid surgery (resections, disinsertions with transpositions of lifting muscles of the upper eyelid). It is important to note that any other eye abnormalities, such as strabismus or amblyopia, must be corrected before surgery.

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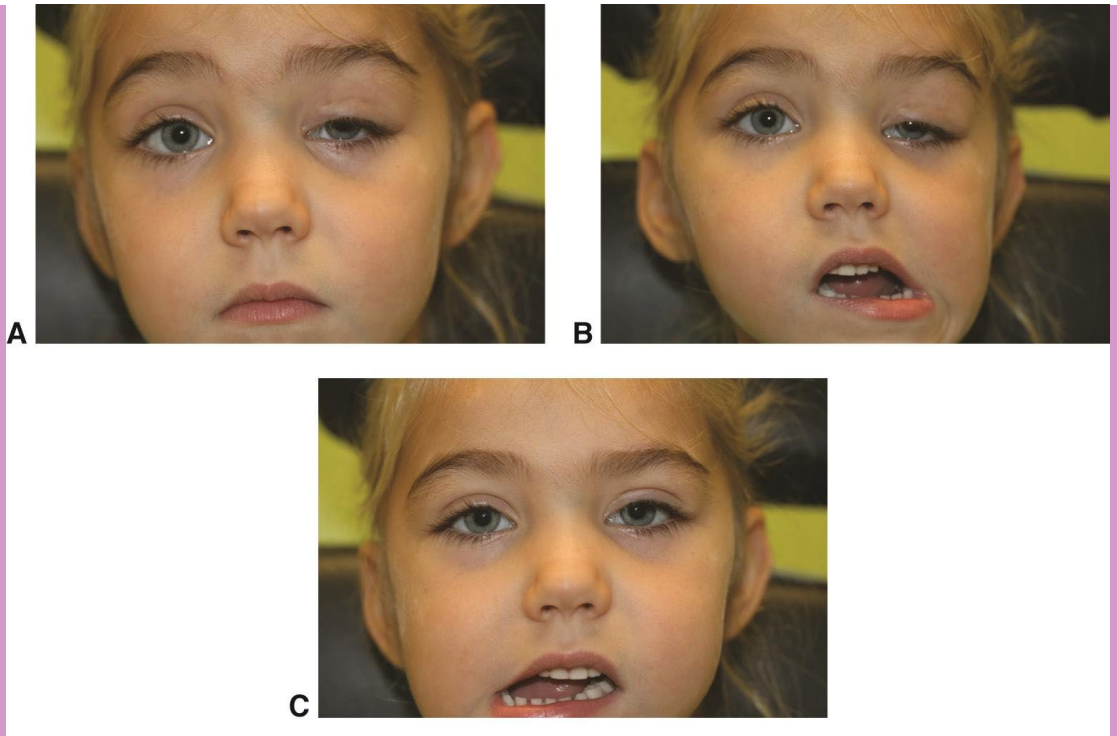
→ **Possible postoperative complications** after eyelid surgery are:

- asymmetry between the two eyelids
- an exaggeration of the retraction movements of the eyelid during chewing, swallowing, etc.
- wound infection
- bleeding and / or hematoma
- granuloma

PROGNOSIS

→ Marcus Gunn syndrome does not significantly affect the patient's quality of life, the consequences being mainly aesthetic (eyelid ptosis).

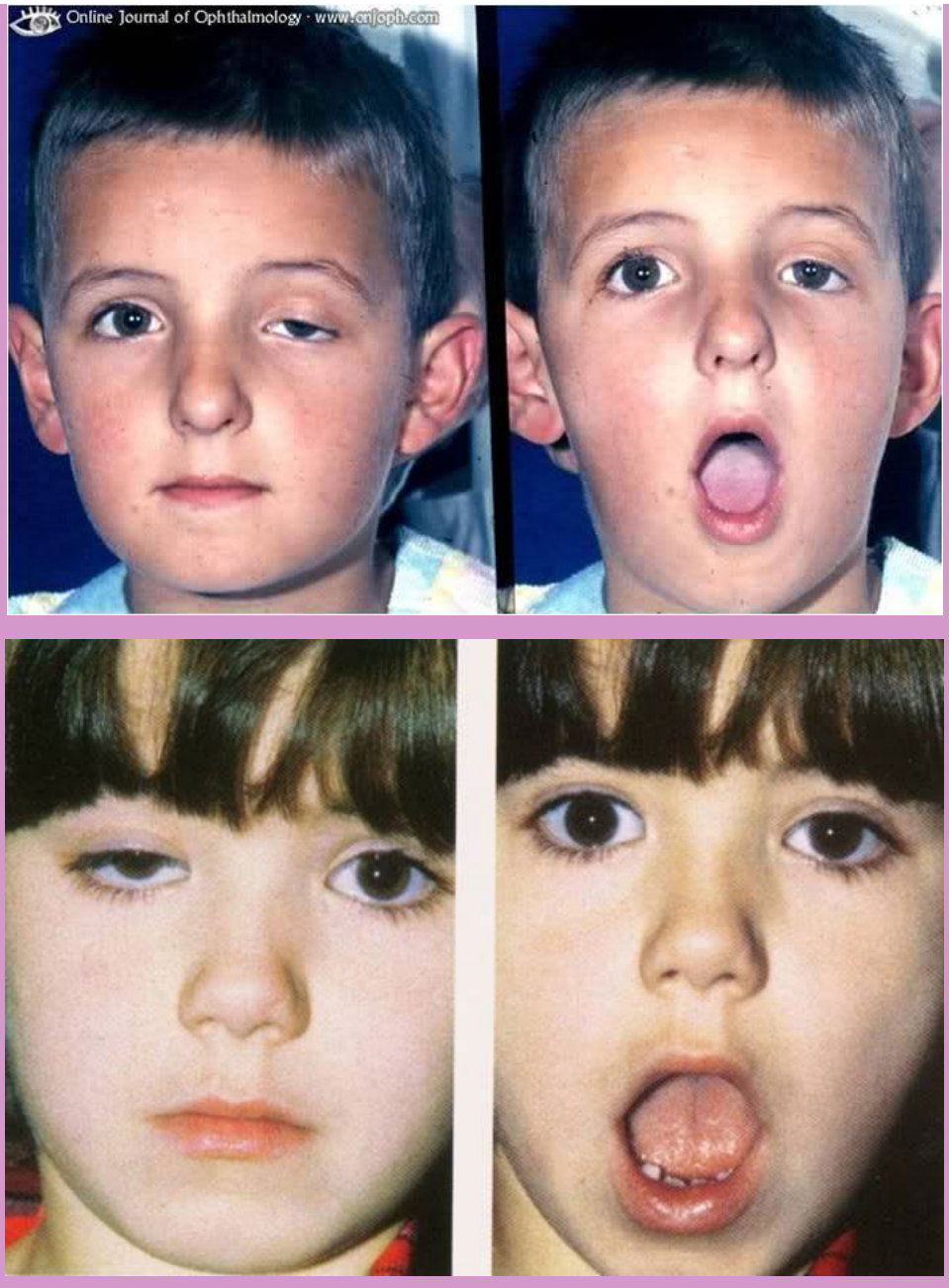
→ Due to a possible association with other eye conditions, such as **strabismus, anisometropia, amblyopia,** a regular eye consultation is recommended.



Img. 1: Marcus-Gunn Phenomenon – correction of ptosis by lateral movements of the mandible

Source: https://www.aao.org/image/marcus-gunn-jaw-winking-syndrome-2

Translation from Romanian



Img. 2: Marcus Gunn Phenomenon – correction of ptosis by opening the mouth

Source:
https://www.atlasophthalmology.net/photo.jsf?node=3920&locale=deDies

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I, BURA TEODORA - NATALIA, certified interpreter and translator for English, by virtue of the authorization no. 35530/20.03.2013, issued by the Ministry of Justice of Romania, hereby certify the accuracy of the translation from Romanian into English.

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