СИНДРОМ НА MARCUS GUNN БЕЗ ПТОЗА

Невяна Велева

Очна клиника, УМБАЛ „Александровска”
Катедра по Офталмология, Медицински Университет-София

MARCUS GUNN SYNDROME WITHOUT PTOSIS

Nevyana Veleva

Ophthalmology Clinic, Alexandrovskia University Hospital
Department of Ophthalmology, Medical University-Sofia

РЕЗЮМЕ

Синдромът на Marcus Gunn е рядко конгенитално заболяване, описано за първи път през 1883г. от Robert Marcus Gunn. При класическата форма на феномена се наблюдава повдигане или ретракция на птозиращата горна клепач при движение на долната челюст. В литературата са описват и редки форми на синдрома, при които не се наблюдава птоза. Представен е рядък клиничен случай на 8-месечно бебе, при което е диагностициран феноменът на Marcus Gunn при липсата на конгенитална птоза.

Ключови думи: синдром на Marcus Gunn, рядък случай, без птоза

ABSTRACT

Marcus Gunn syndrome is a rare congenital disease, first reported by Robert Marcus Gunn in 1883. Classically, the phenomenon is characterized by ptotic eyelid elevation or retraction with jaw movements. In literature, rare forms of the syndrome without ptosis have been discussed. We report a rare clinical case of 8-month-old baby who was diagnosed with the Marcus Gunn phenomenon without congenital ptosis.

Keywords: Marcus Gunn syndrome, rare presentation, without ptosis
ВЪВЕДЕНИЕ
Синдромът на Marcus Gunn, или т.нар. “jaw-winking” феномен, е рядко конгенитално заболяване, описано за първи път през 1883 г. от Robert Marcus Gunn (1). Обозначава се още като тригемино-окуломоторна синкенезия или птериго-леваторна синкенезия и е най-честата форма на конгенитална синкенетична птоза (2). Синдромът се характеризира с наличието на конгенитална птоза на горния клепач и синкинетично повдигане и дори ретракция на засегнатия клепач под формата на примигване при движение на долната челюст (3). По последни литературни данни синдромът на Marcus Gunn съставлява 8.5% от всички случаи на конгенитална птоза (4), като за българската популация честотата му е 5.4% (5). По правило феноменът се наблюдава единстрано, макар че са описани и изяви на двуоочна патология (2,6,7).

В повечето случаи се касае за спорадично засягане, като рядко описаните фамилни случаи имат АД механизъм на унаследяване (8). В настоящето изложение е представен рядък вариант на синдрома на Marcus Gunn, при който липсва конгенитална птоза.

КЛИНИЧЕН СЛУЧАЙ
8-месечно бебе от мъжки пол е прегледано в Детски очен кабинет, Очна клиника, УМБАЛ „Александровска“, София, по настояване на майката. Скоро след раждането на детето тя забелязала, „че при сукане горният клепач на лявото око пада надолу, докато горният клепач на дясното око стои повдигнат и детето примигва с него“. Според майката проблемът е в лявото око и по този повод търси очна консултация. Детето е родено на термин от втора нормално протекла бременност, като по време на пренаталната диагностика са установени анормални на бъбречите. Детето е с нормално физическо и нервно-психическо развитие, отговарящи на възрастта.

Налице са конгенитални аномалии на отделителната система, включително двоен бъбрек с хидронефроза вдясно, нефункциониращ ляв бъбрек и двустранно мегауретери. Осъществи се пълен офталмологичен преглед, съобщи сен дебелата от ясността на бъбречите. Детето е с нормално физическо и нервно-психическо развитие, отговарящи на възрастта. Налице са конгенитални аномалии на отделителната система, включително двоен бъбрек с хидронефроза вдясно, нефункциониращ ляв бъбрек и двустранно мегауретери. Осъществи се пълен офталмологичен преглед, съобщи сен дебелата от ясността на бъбречите. Детето е с нормално физическо и нервно-психическо развитие, отговарящи на възрастта.

Инклинационный статус — липса на птоза (Фиг. 1), нормален ортоптичен статус; рефракция в нормите за възрастта, без наличието на птоза (Фиг. 2). Marcus Gunn syndrome without ptosis in right eye was diagnosed due to presence of oculofacial (pterygo-levator) synkinesis in the absence of congenital ptosis. Mother was informed about the child’s disorder and the absence of necessity for treatment at the moment.

INTRODUCTION
Marcus Gunn syndrome or the so-called “jaw-winking” phenomenon is a rare congenital disorder, described for a first time by Robert Marcus Gunn in 1883 (1). It is known as trigemino-oculomotor synkinesis or pterygoid-levator synkinesis and is the most common form of congenital synkinetic ptosis (2). The syndrome is characterized by congenital ptosis and synkinetic elevation and even retraction of the affected eyelid as winking triggered by jaw movement (3). An updated literature data reveal that Marcus Gunn phenomenon is responsible for up to 8.5% of all cases with congenital ptosis (4), with an incidence rate of 5.4% for the Bulgarian population (5). In most cases it is an unilateral disease, although bilateral cases have also been described (2,6,7). It is usually considered a sporadic condition, though familial cases have been documented (8). In this article a rare variant of Marcus Gunn phenomenon without congenital ptosis is presented.

CASE REPORT
An 8-month-old baby boy was examined at Pediatric Ophthalmology Unit, Ophthalmology Clinic, “Alexandrovska” University Hospital, Sofia, Bulgaria. Soon after birth the mother noticed that “during sucking the left upper eyelid drops while right upper eyelid elevates and baby winks with it”, thinking that the problem was with the baby’s left upper eyelid. The boy was born full-term from a second normal pregnancy. During prenatal screening examinations congenital renal anomalies were diagnosed. The baby was with normal physical and mental development appropriate for the age. The congenital renal anomalies were right double kidney with hydronephrosis, left nonfunctional kidney, and bilateral megaureters. Full eye exam, appropriate for the child’s age was performed. No ocular pathology was detected, there was absence of ptosis (Fig. 1); normal orthoptic status; refraction in normal ranges without anisometropia, normal anterior and posterior segments. Afterwards the baby was examined during bottle feeding and rhythmic winking movements with the right eye leading to upper eyelid elevation and widening of the right palpebral fissure on each suction motion were detected (Fig. 2).

Marcus Gunn syndrome without ptosis in right eye was diagnosed due to presence of oculofacial (pterygo-levator) synkinesis in the absence of congenital ptosis. Mother was informed about the child’s disorder and the absence of necessity for treatment at the moment.
DISCUSSION

Symptom severity in Marcus Gunn syndrome can vary significantly — from barely visible defects to significant cosmetic and functional disorders. Variations can be observed not just in ptosis grade — mild, moderate and severe, but in the magnitude of jaw-winking — from mild (< 2 mm) and moderate (2-5 mm) elevation to severe eyelid retraction (≥ 5 mm) triggered by jaw movement (9). The elevation of the affected eyelid is triggered by chewing, suction, lateral mandible movement, smiling, but also by sternocleidomastoid contraction, protrusion of the tongue, Valsalva maneuver, and even by breathing (10). It is evident at birth and parents are the first that detect the jaw-winking during infants’ feeding (3).

Different theories have been discussed regarding the etiology and pathogenesis of the phenomenon. Classic theory denotes anomalous connections between the mandibular branch of the trigeminal nerve and the superior branch of the oculomotor nerve (10). Thus the upper eyelid levator receives double innervation. A second theory is the hypothesis about the central origin of the syndrome. It discusses the presence of aberrant connections in the midbrain between the trigeminal nucleus and the oculomotor nucleus (2), or the neural misdirection of the trigeminal motor axons of the upper eyelid elevator (8). A third theory about the atavistic origin has also been discussed in the literature (3). It is based on the fact that in some lower animals and fishes the retraction of the upper eyelid is associated with opening of the mouth.

Patients often have associated ophthalmological abnormalities including amblyopia, strabismus, anisometropia, paralysis of the rectus superior, double elevator palsy (3,10). Rarely, associated systemic anomalies such as congenital pathology of the extremities, spina bifida, renal calculi and congenital renal anomalies, cleft lips, etc. are reported.

Syndrome treatment is conservative and surgical. Conservative therapy includes refractive error and anisometropia optical correction, ambioplia treatment. Surgical therapy aims to correct both, jaw-winking synkinesis and ptosis (9), but before surgery is initiated two facts must be taken into account. First, literature review shows opposing views for ptosis evolution, with some authors declare that with child’s growth reduction in ptosis severity can be observed (3). Second, with time the child will go to recognize which movements are responsible for the synkinesis and will learn how to control them, thus minimizing or masking the syndrome (11).

The presented clinical case is an extremely rare variant of the Marcus Gunn syndrome, characterized by trigemino-oculomotor synkinesis in the absence of congenital ptosis. Some other cases of the jaw-winking phenomenon without congenital ptosis are published in medical literature (4,12,13), outlining that this form is responsible for just 5.6% of all Marcus Gunn syndrome cases (4). Nevertheless, some authors’ opinion that this low rate can be explained mainly with the low pick up rate due to nondiagnosis by medical specialists because of ptosis absence and unfamiliarity with this variant or unsought medical consultation (14). It is undisputed that Marcus Gunn syndrome without ptosis is an extremely rare clinical finding and an interesting clinical example of the variability of different symptoms in the context of one syndrome.
Marcus Gunn Syndrome Without Ptosis

REFERENCES