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Case report

# Three-year-old child with meroacrania – Neurological signs

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**Abstract**

Neurological findings in a three-year-old child with meroacrania provide new insights into how the nervous system develops and functions in the absence of superior levels of control from the time of origin. The girl is the first child of a non-consanguineous white Brazilian couple, born at term, weighing 2650 g and measuring 44 cm in length. Upon examination at 43 months, she had quadriplegia, global hypotonia with occasional body hypertonia in a decorticate posture, hyperreflexia, ankle clonus, and extensor plantar response. This case allowed us to verify that, in the absence of upper structures and subcortical nuclei, there are clear signs that suggest corticospinal primacy in motor functions without a substitute pathway. Sound orientation responses suggest the independence of the vestibular-acoustic-ocular system, and manifestations of responsiveness to the environment raise questions about consciousness.


*Keywords:* Anencephaly; Child; Assessment; Primitive reflexes; Motor control

## 1. Introduction

Meroacrania is a kind of anencephaly wherein some vegetative functions are preserved, allowing longer life span [1]. There are few publications about neurological semiology in infants with meroacrania [2]. Thus, the objective here is to discuss the neurological findings from a 43-month-old child with meroacrania.

## 2. Case report

The patient is the first child of a healthy non-consanguineous, Brazilian white couple. The pregnancy came to term without complications, and the newborn was 2650 g and 44 cm in length at birth. No malformation was detected except for a small sac with scattered nonviable neural tissue in the vertex. A cranial X-ray suggested meroacrania, since the occipital bone and foramen magnum were present. She had plastic surgery in the vertex and left the hospital on the 15th day, with no oxygen support and no need for a feeding tube.

At 43 months of age, her weight and height are under the 3rd percentile; her heart rate is around 50 beats/min, she experiences short apneas and difficulty in swallowing (the parents refused a gastrostomy), as well as urinary retention once a month and severe constipation. Her body temperature varies according to the environment. She responds to pain by muscle contractions or feeble crying. She smiles upon sound or tactile stimulus as well as while sleeping. Dentition is appropriate for her age. Upon examination in the supine position, she presents

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global hypotonia, rare proximal movements only in upper limbs, inactive hands, and adducted thumbs. Without external stimulation, there are rare spasms in the decorticate posture and she showed smooth ocular movements, sometimes conjugate or dissociate, without tremor or nystagmus. The pupils are 2 mm to the left and 1 mm to the right. Photomotor and consensual reflexes are slow to the left, and absent to the right. Fundus oculi examination was possible only on the left side and presents regular vessel distribution, an apparently normal retina and a small pinkish papilla. There is hypomimia with weak but complete eyelid closure. Oculocephalic reflex is slow. Corneal-palpebral, sternutatory, glabellar, mentum and orbicular of lip reflexes are tenuous, as well as bilateral palmomental reflex. She has good apposition of the lips and no sialorrhea; sucking is present and the rooting reflex is absent.

She simultaneously moves her head and eyes towards a sound source at the horizontal level. Upon a cochlear-palpebral reflex test she responds with whole-body startle, arm abduction with no hand opening or adduction. With respect to the Moro reflex, she does not respond to the André-Thomas maneuver when we cause her head to fall in a dorsal position. Palmar and plantar grasps are absent. Asymmetric and symmetric tonic neck reflexes show fragmentary responses only in superior limbs and tonic labirintic reflex is absent. When given trigeminal stimulation at the nose while sitting on her calves with knees flexed, she extends her neck and trunk. The same response occurs when in the prone position (Fig. 1). She changes from ventral to the side position in block; however, this righting reflex does not happen in the supine position. A sustained Landau's reflex is present. There is no stepping reflex, even with extension of the head. There is no placing reflex. Supporting reaction is present, extending the lower limbs only. During Bobath's maneuver (suspension vertically, no foot support, held by armpit) lower limbs look hypotonic. However, the child crosses them (scissors) when there is a hyperextension of the spine by forcing shoulders back. In the prone position, only hypotonia is present at rest, without the intermittent spasms as seen in the supine position, and spontaneous crawling reflex appears intensified with Bauer's maneuver by offering a plantar support. She does not present either lateral support or parachute for both upper and lower limbs.

While testing tone by knee flexo-extension, at first a clasp-knife appears, with no resistance in the sequence. There is spontaneous ankle clonus and, when tested, there are more than 10 beats. Hoffman's sign is obtained bilaterally. Dorsiflexion of the hallux is spontaneous and increases without fanning the other toes by Babinski's classical test and Gordon's, Chaddock's, Moniz's, Bing's and Gonda's technique variants. There is no reaction in triple flexion, or in the crossed extensor reflex. All stretch reflexes are hyperactive. There are no responses to abdominal cutaneous reflexes, trunk bending or Galant's reflex. The patient exhibits Gamper's sign (Fig. 2): in supine position, her lower limbs are pressed in extension against the examining table and in the sequence she flexes her trunk (bowing reflex).

Brain magnetic resonance imaging at 14 months of age (Fig. 3) showed thalamus, pituitary gland and optic nerves, mesencephalon, vermis and cerebellar hemispheres and no telencephalon; reduced brainstem volume; the fourth ventricle was verticalized.

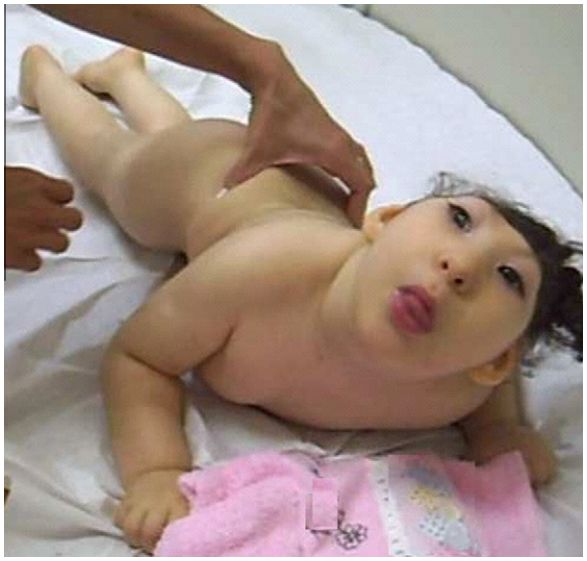

Fig. 1. Extension of the neck and trunk after touching the nose upward (trigeminal stimulation).

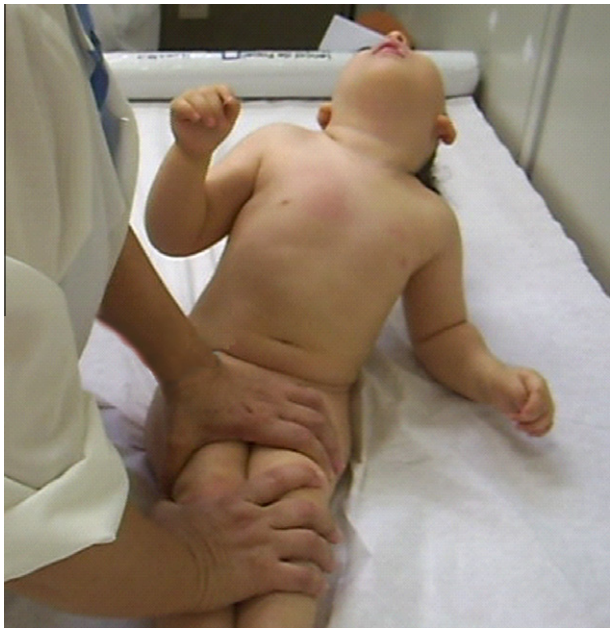

Fig. 2. Gamper's sign, pressing lower limbs, in a three-year-old anencephalic girl.

## 3. Discussion

A neurological exam of this anencephalic girl at 3 years of age raises some issues related to the hierarchical processes of the nervous system function in humans.

Her motor findings are similar to a corticospinal deficit [3]. In acute stages of corticospinal lesions, hypotonia in limbs is present, with progression to spasticity in different timings, as commonly seen in neuroclinics and described by experimental studies [4,5]. However, for unexplained reasons, there are patients in which hypertonia does not come about. Our patient shows brief periods of hypertonia, like spasms. This peculiar find was described during the neonatal period in a similar case of meroacrania [2]. Therefore, it seems that there is an arrest on maturation of the pathways related to control of tonus; prevailing those related to the hypotonia, but with an intermittent firing of a decorticate state. Concerning the extensor plantar response, although her persistent hallux dorsiflexion could represent a pseudo-Babinski sign [6], this is less likely than a pyramidal sign, considering the absence of extrapyramidal signs, which is expected at least in the upper limbs that exhibit movements and because of the marked hypotonia.

Another peculiar finding is the good muscle trophism, despite a three-year-paralysis. A normal morphology of the muscles of an anencephalic fetus was described, raising speculations on this intriguing finding [7]. The spinal cord of our patient is provided with motoneurons, since there are phasic reflexes, but there is no corticospinal influence, resulting in paralysis, thus, we can speculate that either the muscle has inner mechanisms to maintain its trophism, despite the absence of movement, or there are particular types of motor unit and neuromuscular junction in meroacrania.

The persistence of some primitive reflexes in this girl can be explained by the absence of cortical inhibition [8]. Primitive reflexes were found weak or absent in neonates with meroacrania at different levels of commitment [2]. The patient 1 of that series [2]; with no brain tissue above foramen magnum and expired around 11 h after birth, had no sucking reflex, and slight palmar, plantar grasps, Moro reflexes and bilateral ankle jerks could be elicited. The presence of grasping and Moro reflexes in the neonatal period and the onward absence as in our patient, at the age of 43 months might suggest a role of unidentified paths from the brainstem to the lower motor neurons in the establishment of inhibition on the primitive reflexes during development. On the other side, the absence of sucking reflex, as in the neonate with meroacrania [2], occurs in many other conditions with different levels of brain lesions. The presence of sucking reflex in our patient, as we mentioned before, could be due to lack of cortical inhibition, producing perseverant movements, in this case with preservation of cranial nerves involved in the sucking. Stepping reflex has been cited as proof that the spinal cord is a site of gait generators in humans [9]. In spite of the fact that gait generators are present in the spinal cords of cats [10], this has not been demonstrated in humans [11]. Our patient did not present stepping reflex at 43 months of age.

Interesting was the presence of supporting reaction, that is considered resulting of a proprioceptive feedback, by stimulating the limbs and axial extensor muscles with plantar support [8]. We suggest that the Gamper reflex is the result of a very low threshold for a righting reaction, since uprighting the trunk occurs when the lower limbs are forced to extend in a dorsal position. The same reasoning could be used with respect to the influence of trigeminal stimulation and the extensor response of the neck and trunk in a prone position, as presented in Fig. 1. The presence of Landau reflex suggests that the emergence of this reflex does not depend on the maturation of connections to telencephalic structures. The head turning to a sound stimulus addresses the independence

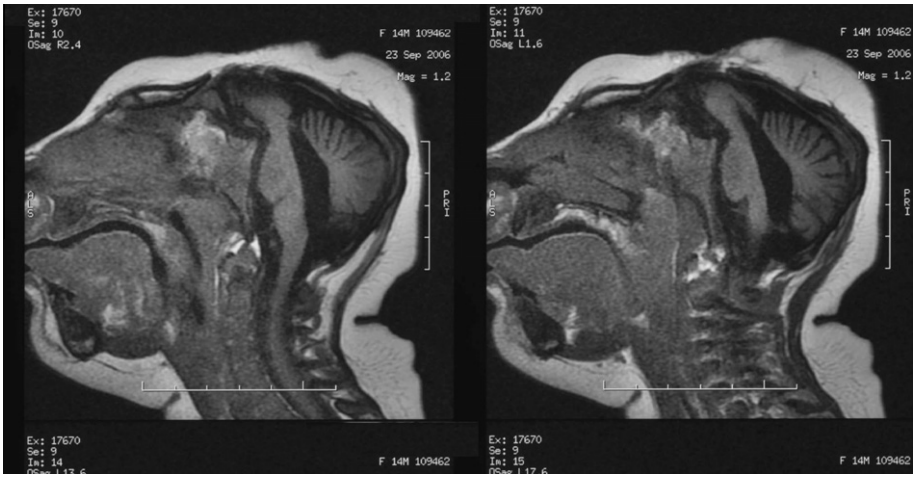


Fig. 3. Sagittal FLAIR magnetic resonance images showing the brainstem and cerebellar structures.

of the acoustic-vestibular path, apart from voluntary movement.

Since Gamper's report [12], smiling has been recognized in anencephalic infants and is related to the rapid eye movement (REM) phase of sleep, which correlates anatomically to the pontine tegmentum. Smiling during pleasurable moments is probably the result of sensitive or sensorial pathways and facial nuclei connections in brainstem [13]. It is noteworthy that this patient had no laughter, expression that requires higher brain functions [14].

Anencephaly raises many questions on functional aspects of the brainstem, cerebellum and spinal cord without supratentorial connections, and in the absence of brain cortex, about consciousness.

Consciousness is regarded as a state or activity characterized by the integration of complex components such as sensation, emotion and intention [15]. Therefore, in children with no brain hemispheres, reactions such as crying and smiling, as well as motor and autonomic responses to stimuli might be regarded as signs of consciousness that lack intention or thinking domains.

## Appendix A. Supplementary data

Supplementary data associated with this article can be found, in the online version, at doi:10.1016/j.braindev.2010.02.001.

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