Mystery Case: Pontine tegmental cap dysplasia in a neonate

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Neurology® 2018;91:e2100-e2101. doi:10.1212/WNL.0000000000006578

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Figure 1 Cranial MRI of the neonate

Midsagittal T2-weighted image (repetition time/echo time 3,600/100 ms) of the patient at her 13th day shows flattening of the ventral pons and dysmorphism of the dorsal upper pons (caplike) protruding in the fourth ventricle (white arrow).

A 2-month-old girl with swallowing difficulty initiated at day 1 was evaluated. Birth history was unremarkable. Her suck, Moro, and grasp reflexes were normal; however, she could not swallow. She had soft palate sagging. She had hearing impairment in her right ear. Basic biochemistry, metabolic tests, and karyotype and microarray studies were normal.

A cranial MRI on day 13 showed tegmental cap (figures 1 and 2). Tegmental cap is a congenital hind brain anomaly with developmental delay, oculomotor disturbances, facial involvement, sensorineural deafness, and swallowing difficulty. Neuroimaging is diagnostic with flat ventral pons, an abnormal curved structure of the pontine tegmentum projecting into the fourth ventricle, and varying degrees of cerebellar peduncle anomalies. Failure of axonal guidance in utero is the most likely pathophysiology.^{1,2}

Author contributions

N. Bilgin performed acquisition of data. Ş. Parlak performed analysis and interpretation. P.Ö. Şimşek-Kiper performed acquisition of data. M. Topcu performed study supervision. H. Topaloglu performed critical revision of the manuscript.

Study funding

No targeted funding reported.

Survey and results

[NPub.org/mc9122](https://www.ncbi.nlm.nih.gov/pubmed/30111122)

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Go to [Neurology.org/N](https://www.neurology.org/N) for full disclosures. Funding information and disclosures deemed relevant by the authors, if any, are provided at the end of the article.

Figure 2 Cranial MRI of the neonate

Axial T2-weighted image at the level of midpons. The white star indicates asymmetrically thin right side of the pons. White arrow shows the tegmental cap.

Disclosure

The authors report no disclosures relevant to the manuscript. Go to [Neurology.org/N](https://www.neurology.org/N) for full disclosures.

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Mystery Case responses: A 2-month-old girl with difficulty swallowing

The Mystery Case series was initiated by the *Neurology*[®] Resident & Fellow Section to develop the clinical reasoning skills of trainees. Residency programs, medical student preceptors, and individuals were invited to use this Mystery Case as an educational tool. Responses to multiple choice questions formulated using this case were solicited through a group email sent to the American Academy of Neurology Consortium of Neurology Residents and Fellows and through social media. We received 145 responses. The majority of respondents

(75%) had been in practice for 1–4 years; 57% were residents or fellows and 31% were faculty/board-certified physicians; the remainder were medical students or advanced practice providers. Sixty-two percent resided outside the United States. A wide range of practice settings was represented.

When presented with this 2-month-old patient's history and examination findings and shown the representative T2 MRI brain slices, 39% correctly identified the thin right side of the pons, 26% also identified flattening of the ventral pons, while only 13% also recognized the protrusion of the dorsal pons. The most frequently selected incorrect options were agenesis of the corpus callosum (55%) and of the cerebellar vermis (28%); the cuts presented were perhaps not ideal for visualizing these structures, but these were not the evident abnormalities.

Upon then being asked to select the most likely diagnosis based on these findings, 16% correctly selected pontine tegmental cap dysplasia. The most popular but incorrect choices were olivopontocerebellar atrophy (19%) and pontocerebellar hypoplasia type 2 (17%). Pontocerebellar hypoplasia is a group of progressive degenerative diseases that begin prenatally and continue postnatally and involve not only the cerebellar system but also have extensive cortical and basal ganglionic abnormalities even in motor neurons, causing a clinical presentation at birth that can resemble spinal muscular atrophy.¹ Olivopontocerebellar hypoplasia, on the other hand, is not a developmental disorder of the nervous system but rather a form of multisystem atrophy characterized by predominant cerebellar features.

This pediatric neurology case highlights an important developmental abnormality of the nervous system and also demonstrates the value of carefully reviewing midline structures on neuroimaging.

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This information is current as of November 26, 2018

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