
Neuroimaging Highlight

Editor: David Pelz

Iniencephaly in an Adult Patient

Submitted by: Martin Côté, André Turmel, Karine Michaud, Pascale Lavoie, Jacques Francoeur, Claude Picard

Can J Neurol Sci. 2012; 39: 385-387

A 24-year-old woman presented to the emergency department with a history of headache and malaise. She was known for a cervico-occipital meningoencephalocele repair and the placement of a ventriculoperitoneal (VP) shunt in infancy; she has otherwise been developing normally, has a normal intellect and is currently living independently. She had a short neck with low posterior hairline, and obvious spinal deformity (Figure 1). Neurologic examination was unremarkable with absence of long tract signs, no gait abnormality, no oculomotor or other cranial nerve deficits and no evidence of gross cerebellar dysfunction. Maternal and family history were negative. Her VP shunt had been revised six weeks earlier following an episode of malfunction. Investigation revealed an infection of the VP shunt with *Propionibacterium acnes* (*P. acnes*); the shunt hardware was therefore removed, an external ventricular drain was installed and she was placed on intravenous antibiotic therapy. After an appropriate course of antibiotics, magnetic resonance imaging (MRI) was done to assess the possibility of an endoscopic third ventriculostomy (ETV) (Figure 2). Judging the situation potentially favourable, we performed an ETV and the external ventricular drain was left in place. However, the patient could not be weaned from her ventricular drain, so a new VP shunt was installed after confirmation that her cerebrospinal fluid (CSF) was sterile. The postoperative course was uneventful, and she was discharged three days after surgery. At three months follow-up, she is currently doing very well.

DISCUSSION

Iniencephaly is a rare neural tube defect characterized by retroflexion of the head, an occipital bone defect and rachischisis of the cervical and thoracic spine. It can be associated with other abnormalities of the central nervous system such as anencephaly, encephalocele and hydrocephalus, as well as malformations of other organ systems¹. The prognosis of affected subjects is usually dismal, with only eight living cases reported in the literature, aged between two months and 17 years old²⁻⁷. It is hence very uncommon to assess the imaging features of iniencephaly in an adult patient.



Figure 1: Physical appearance of our patient.

In our patient, all three cardinal features of iniencephaly are found. On the T2-weighted sagittal MRI, fusion of cervical vertebrae along with severe lordosis of the cervical spine is clearly visible (Figure 2). The head computed tomogram (CT) scan shows an enlarged foramen magnum with a severely hypoplastic posterior fossa, which at birth was associated with a cerebellar meningoencephalocele (Figure 3). The failure of

From the Department of Neurosurgery, Centre Hospitalier Affilié Universitaire de Québec, Hôpital de l'Enfant-Jésus, Université Laval, Québec, Canada.

RECEIVED MAY 11, 2011. FINAL REVISIONS SUBMITTED NOVEMBER 28, 2011.

Correspondence to: Martin Côté, 1401, 18^e rue, Québec, QC, G1J 1Z4, Canada. Email: martin.cote.19@ulaval.ca

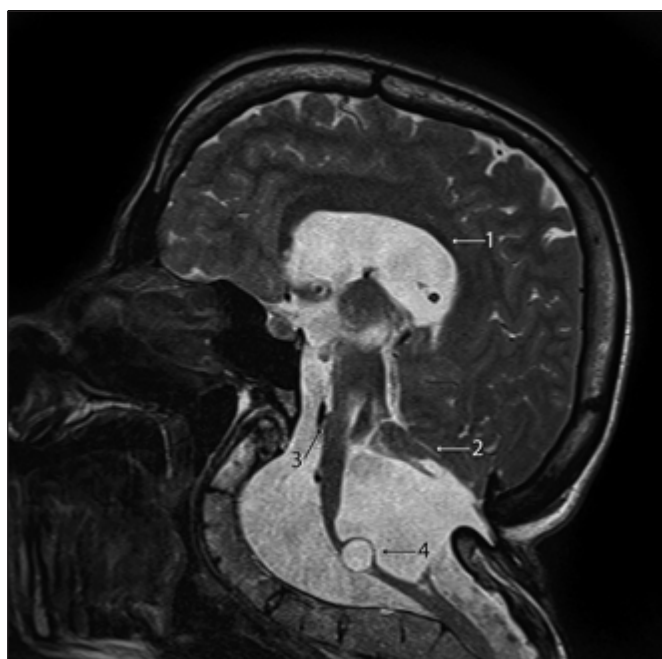


Figure 2: T2-weighted sagittal MRI of the head showing severe lordosis of the cervical spine with fusion of vertebral bodies. Also apparent are (1) callosal dysgenesis, (2) cerebellar hypoplasia, (3) brain stem malformation and (4) an intramedullary cyst.



Figure 3: CT-scan of the foramen magnum revealing an enlarged foramen magnum.



Figure 3: CT scan of the cervical spine at C5 level showing failure of fusion of the posterior elements.

fusion of the posterior elements of the cervical spine, seen on the cervical CT scan (Figure 4), completes the characteristic triad of this disease⁴. Other abnormalities observed on the MRI of our patient, all known to be related to iniencephaly⁸, include corpus callosum dysgenesis, cerebellar hypoplasia, brain stem malformation and shunt-dependent hydrocephalus. Moreover, an intramedullary inclusion cyst can be seen in the cervical spinal cord. Since diffusion weighted imaging failed to show restricted diffusion, we believe that this is a neuroglial or neurenteric cyst, known to exist with the Klippel-Feil malformation⁹, although in the context of iniencephaly, such lesions have been described in the cerebellum but not in other locations of the central nervous system⁸.

The embryonic origin of iniencephaly is speculative, but some attribute a primary defect of segmentation of neural tube formation as a possible mechanism for its happening. The segmentation at the occipito-cervical level is under the control of Hox homeobox family of genes. Mutations in Hox genes would be responsible for the anomalies of the neural tube and the mesodermal defects of the basiocciput and high cervical spinal column¹⁰⁻¹³. The association of the Klippel-Feil anomaly in our patient could be explained by Hox homeobox family mutations, but also could implicate the Pax-1 gene expression, resulting in vertebral fusions and other notochordal defects, such as inclusion cysts⁸.

CONCLUSION

To the best of our knowledge, this is the first reported case of a patient with iniencephaly surviving into adulthood. In accordance with the published literature, imaging features in our patient consist of an occipital defect, severe retroflexion of the cervical spine, rachischisis of the cervical spine, and other abnormalities of the spine and central nervous system. Mutations in Hox homeobox family of genes and problems with Pax-1 gene expression could be at the origin of this very unusual malformation.

REFERENCES

1. Joo JG, Beke A, Papp C, Szigeti Z, Csaba A, Papp Z. Major diagnostic and pathological features of iniencephaly based on twenty-four cases. *Fetal Diagn Ther*. 2008;24:1-6.
2. Basu S, Muthusami S, Bhutada A, Kumar A. Iniencephaly: association with unusual congenital malformations. *Ind J Ped*. 2008;75:1189-90.
3. Aytar MH, Dogulu F, Cemil B, Ergun E, Kurt G, Baykaner K. Iniencephaly and long-term-survival: a rare case report. *Childs Nerv Syst*. 2007;23:719-21.
4. Erdinciler P, Kaynar MY, Canbaz B, Kocer N, Kunday C, Ciplak N. Iniencephaly: neuradiological and surgical features. *J Neurosurg*. 1998;89:317-20.
5. Munden MM, Macpherson RI, Cure J. Iniencephaly: 3D-computed imaging. *Pediatr Radiol*. 1993;23:572.
6. Katz VL, Aylsworth AS, Albright SG. Iniencephaly is not uniformly fatal. *Prenat Diagn*. 1989;9:595-9.
7. Sherk HH, Shut L, Chung S. Iniencephalic deformity of the cervical spine with Klippel-Feil anomalies and congenital elevation of the scapula. *J Bone Joint Surg (Am)*. 1974;56:1254-9.
8. Jeanne-Pasquier C, Carles D, Alberti EM, Jacob B. Iniencephalie: à propos de 4 nouvelles observations et revue de la littérature. *J Gynecol Obstet Biol Reprod*. 2002;31:276-82.
9. David KM, Thorogood PV, Stevens JM, Crockard HA. The dysmorphic cervical spine in Klippel-Feil Syndrome: interpretations from developmental biology. *Neurosurg Focus*. 1999;6:e1
10. Sarnat HB, Menkes JH. The new neuroembryology: How to construct a neural tube. *J Child Neurol*. 2000;21:109-24.
11. Tümpel S, Wiedemann LM, Krumlauf R. Hox genes and segmentation of the vertebrate hindbrain. *Curr Top Dev Biol*. 2009;88:103-37.
12. Wellik DM. Hox genes and vertebrate axial pattern. *Curr Top Dev Biol*. 2009;88:257-78.
13. Wellik DM. Hox patterning of the vertebrate axial skeleton. *Dev Dyn*. 2007; 236: 2454-63.