Cerebellar tonsil herniation: Its diverse pathogenesis

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Authors: H. Mukai, H. Yokota, T. Horikoshi, K. Motoori, T. Uno; Chiba/JP
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Learning objectives

The causes of cerebellar tonsil herniation (CTH) are very different and the clinical outcomes are varied.

Congenital causes include hypoplasia of the posterior fossa, acquired causes include increased volume of the intracranial contents and abnormal intracranial pressure.

CTH is not rare. Interpreting image characteristics and pathogenesis makes understanding clinical significance on each disorder with CTH.

We propose to attend this objectives:

• illustrate a summary of CTH.
• illustrate development of the skull related with CTH on embryologic aspect.
• present the concrete examples of some diseases associated with CTH.
Background

1. What is the cerebellar tonsil herniation?

Cerebellar tonsil herniation (CTH) is called as the tonsillar herniation, the downward cerebellar herniation and the transforaminal herniation.

Depending on the degree of retraction, the degrees of symptoms also vary from none to serious.

With wide foramen magnum or slowly progressive situation, it is only accompanied by slight symptoms or sometimes no symptoms.

CTH may be accompanied by increased intracranial pressure and myelopathy associated with syringomyelia, may also require the decompression by the surgery.

Due to retraction of the medulla oblongata, impairment of respiratory center induces ataxic breathing and respiratory arrest. CTH may cause disturbance of consciousness after apnea. CTH may be lethal condition.

The tonsils more than 5mm below the line connecting with the lower pole of the clivus and the occipital bone are considered abnormal (Fig.1). Some authors report that CTH is diagnosed by tonsils more than 3mm below the foramen magnum because of higher sensitivity, but it may be observed 3-5mm descent in healthy group.

The physiological range of descent of the tonsils is 6mm or less in 10-year-old or younger and 5mm in from 11 to 33-year-old. Seventy percents of the patients with 5-10mm below the foramen magnum and nearly all of patients with 12mm below it, have symptoms.

2. What causes the cerebellar tonsil herniation?

Causes of the CTH are mostly disproportion between volume of the intracranial contents and skull structure, the other causes are mechanical or functional factors accompanied with the pressure gradient above and below the foramen magnum. (table.1)

Increased volume of intracranial contents include megalencephaly, hydrocephalus and space-occupying lesions such as tumors or hemorrhage.

The structural abnormalities of the skull induced CTH is a decrease of the intracranial volume, in particular a decrease of the volume of the posterior fossa associated with malformations or developmental disabilities. These are mostly congenital pathology. Malformations include Chiari malformation, and developmental disabilities include skeletal dysplasia leading to craniosynostosis such as Crouzon disease.
Functional factors such as intracranial hypotension by spontaneous or secondary cerebrospinal fluid leakage and increased intracranial pressure, may cause CTH.

3. Development of the posterior fossa

In order to understand dysplasia of the posterior fossa, it is necessary to understand the development of it.

Most of bones of the trunk is derived from mesoderm. Skull including the frontal bone and facial bones are derived from the neural crest. In contrast parietal bone and occipital bone, petrous portion of the temporal bone are derived from the paraxial mesoderm. (fig.2)

The cranium can be developmentally divided into neurocranium to protect the brain and viscerocranium to form the skeleton of the face.
I will describe with respect to the neurocranium involved in the development of the posterior fossa.
In the classification of ossification, neurocranium is further divided into cartilaginous neurocranium and membranous neurocranium.

• cartilaginous neurocranium

It consists of cartilaginous primordia of the skull base, bones of the skull base is formed by endochondral ossification.
The occipital bone, the sphenoid bone and the ethmoid is in order ossified. Parachordal cartilage or basal plate is formed around the cephalic end of the notochord, these fuse with cartilage from sclerotome of the occipital somites. The cartilage mass involves in the formation of a basilar portion of the occipital bone, extends while surrounding the cephalic end of the spinal cord and forms a peripheral portion of the foramen magnum.

• membranous neurocranium

Intramembranous ossification of the neural crest mesenchyme at the top and lateral portion of the brain leads to form calvaria.

This means that occipital bone is derived from the paraxial mesoderm unlike the frontal bone and is formed by endochondral ossification unlike the frontal bone and the parietal bone. So these abnormalities causes defective development of the posterior fossa. It can be said that the occipital bone is distinctive among bones of the skull.

There is fibrous sutures in the calvaria of newborn, so the brain volume can increase through in infancy and childhood.
The volume of the calvaria increases the most until 2-year-old, the brain volume increases rapidly from 2 to 5-year-old. Calvaria can extend until 16-year-old. By thickening of the skull for 3-4 years after 16-year-old, calvaria can extend slightly further.

3.5. Does the abnormality of the enchondral ossification cause the cerebellar tonsil herniation?

Abnormality of the endochondral ossification inhibits the development of the posterior fossa including skull base.

Achondroplasia representing abnormality of the endochondral ossification is accompanied not only with hypoplasia of the posterior fossa, but also with narrowing of the foramen magnum at high rates. (fig.3)

Achondroplasia has a direct compression of the medulla oblongata rather than descent of the cerebellar tonsil in many cases.

However, some case accompanied with CTH have been reported.

It often merge hydrocephalus, but not severe.

4. Assessment of the posterior fossa

Several indicators for the evaluation of the posterior fossa have been considered.

Compared with healthy group and Chiari malformation type I, Remy et al. discussed with respect to the neural and the bone structure of the posterior fossa. It have been showed a trend of shorter clivus, shorter supraocciput, smaller midsagittal area of the posterior fossa and increased tentorial angle (normal degree from 27 to 52°). There have been a significant difference in the length of the clivus. (fig.4)

There are some reports that the volume of the posterior fossa is not correlated with CTH.
Fig. 1: 53 years-old female with neuromyelitis optica. She incidentally has 5mm of descent of tonsil. She did not have any symptom due to tonsil herniation.

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Table 1: chart of the causes of CTH

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Fig. 2: Derivation of the bones of the skull.

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Fig. 3: Narrowing of the foramen magnum with achondroplasia

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Fig. 4: Assessment of the posterior fossa

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Findings and procedure details

1) Chiari malformation(Fig.6)

Hans Chiari as Austrian pathologist in 1891 had divided Chiari malformation into four categories:

Type I : more than 5mm of descent of only the cerebellar tonsil

Type II : descent of the cerebellar tonsil, vermis, the fourth ventricle and medulla oblongata

At present, type I and type II are classified by the absence or presence of myelomeningocele

Type III : accompanied with the occipital meningocele

Type IV : cerebellar hypoplasia

At present, this is not included in the Chiari malformation

Hydrocephalus and syringomyelia is accompanied with 10% : 50-85% in type I and 90% : 20% in type II, respectively.

Chiari type I and type II are embryologically distinct disorders.

Chiari type I is defined as the abnormal formation of the paraxial mesoderm, leading to hypoplasia of the occipital bone and the shallow posterior fossa.

Chiari type II is defined as defective development of primitive ventricle (especially near the fourth ventricle) due to discharge of CSF to the amniotic cavity by myelomeningocele, leading to the shallow posterior fossa.

2) Craniosynostosis(Fig7-9)

Craniosynostosis is accompanied with deformity of the skull by the premature fusion of the suture. This incidence rate is one in 2100-2500 people.

Craniosynostosis is mostly correlated with abnormality of TWIST1 gene and fibroblast growth factor receptor(FGFR) gene, such as Apert syndrome, Crouzon disease(Fig.7), Pfeiffer syndrome(Fig.8-9), Muenke syndrome, Beare-Stevenson syndrome and Jackson-Weiss syndrome.
Muenke syndrome and Crouzon disease accompanied by acanthosis nigricans are caused by mutation of the FGFR3, the other diseases are mostly caused by mutation of the FGFR2.

In the report of T. de jong et al, it has been considered that there are no differences between the brain volume in healthy group and in patients with craniosynostosis. Cranioencephalic disproportion makes prone to herniation.

Fusion of lambdoid suture is most correlated with the shallow posterior fossa.

3) Increased volume of intracranial contents(Fig.10)

In many cases, CTH is caused by posterior fossa tumors and cerebellar hemorrhage, and often accompanied with hydrocephalus.

Not only infratentorial lesions but also supratentorial lesions can cause CTH.

3') Acromegaly(Fig.11)

CTH is observed in 15% of patients with acromegaly, it shows more than 5mm of descent of the tonsil in 4.7% in the report of Renzo et al. Some symptoms such as headache and visual disturbance occur. These patients with symptoms are at significantly higher rate than in the healthy group. Those who are lower age, short disease duration, no history of hypertension and no treatment with a growth hormone(GH) receptor antagonist have a tendency to have symptoms. Two cases were accompanied with syringomyelia.

Sievers et al have suggested that GH/IGF-1(insulin-like growth factor) have had a trophic effect of neurogenesis and oligodendrogenesis, therefore that excess secretion of the GH/IGF-1 have caused diffuse increase of extracellular fluid and cell volume.

Overgrowth of bone of the patients with acromegaly is extrovert, so intracranial volume does not correlate to enlarging head circumference. CTH may be improved by treatment for acromegaly, and by brain atrophy when it comes to aging.

4) Abnormal intracranial pressure(Fig.12-13)

Increased intracranial pressure with any causes leads to downward displacement of the intracranial contents to the spinal canal with the relatively lower intraspinal pressure.

Intracranial hypotension/CSF hypovolemia syndrome is a condition of the cerebrospinal fluid leakage occurring with some triggers such as minor trauma and spinal surgery.

CSF leaks causes the downward slope of the pressure above and below the foramen magnum, and induces the descent of the cerebellar tonsils.
Thick dural enhancement, bilateral subdural hematomas, enlarged pituitary gland are sometimes found.

Repetitive lumbar puncture and lumboperitoneal shunt also cause CTH in same mechanism.

Jorge A. Lazoreff et al. have reported CTH induced by long-term ventriculoperitoneal shunt, but have suggested that the patient had been potentially accompanied with hypoplasia of the posterior fossa.

3)+4) Macrocephaly capillary malformation (Macrocephaly-cutis marmorata telangiectatica congenita)(Fig.14)

Macrocephaly-capillary malformation is a multiple congenital anomaly syndrome in children with macrocephaly, overgrowth, skin abnormalities with diffuse marbled skin, philtrum and upper lip vascular anomaly, syndactyly and hyperlaxity of the joint.

It is associated with obstructive/non-obstructive hydrocephalus, and 8-69% of the patients with it is accompanied with CTH.

Hydrocephalus and increased volume of the cerebellum due to edema with the failure of the venous circulation, are considered to cause CTH.
Fig. 5: Chiari malformation

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Fig. 6: Crouzon disease Shallow posterior fossa and CTH are seen on CT

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Fig. 7: Patient suspected Pfeiffer syndrome. Radiohumeral synostosis is seen on radiograph. Midface hypoplasia and shallow posterior fossa is seen on CT and severe CTH on MRI.

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**Fig. 8:** Patient suspected Pfeiffer syndrome. Craniosynostosis involving the lambdoid suture causes shallow posterior fossa and CTH.

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**Fig. 9:** Intracranial space-occupying lesions. Not only infratentorial lesions but also supratentorial lesions can cause CTH.

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Fig. 10: Acromegaly

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Fig. 11: Spontaneous intracranial hypotension Thick dural enhancement, bilateral thin subdural hematoma and enlarged pituitary gland are sometimes found.

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Fig. 12: Hydrocephalus caused by aqueduct stenosis

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Fig. 13: Macrocephaly capillary malformation (Macrocephaly-cutis marmorata telangiectatica congenita) Dilatated lateral ventricle and increased volume of the cerebellum are seen on MRI.

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Conclusion

CTH are very different and the clinical outcomes are varied.

One of the congenital causes is hypoplasia of the posterior fossa. Mesodermal insufficiency induces Chiari malformation type I. Chiari malformation type II is induced by poor development of the primitive ventricle due to discharge of cerebrospinal fluid into the amniotic cavity.

Synostosis of the lambdoid suture causes a shallow posterior fossa on craniosynostoses, for example Crouzon disease.

Acquired causes include mass effect by tumors, intracranial hypertension, lumboperitoneal shunt, idiopathic and postoperative intracranial hypotension.

Their mechanisms can be divided to depression from the posterior fossa and traction from the spinal canal. The causes are not only organic but also functional. It is reported that acromegaly induces CTH due to increased volume of the brain.

CTH is not rare. Interpreting image characteristics and pathogenesis makes understanding clinical significance on each disorder with CTH.
References


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