# Case Report

# Trans-sphenoidal Encephalocele and Craniofacial Dysrhaphism: A Case Report

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***Background:*** *Basal encephalocele is a rare entity of midline neural tube defect. Trans-sphenoidal encephalocele (TSE) is a type of basal encephalocele protruding through the sphenoid bone.*
***Objective:*** *To report clinical manifestation, radiographic findings and surgical treatment in a patient with TSE.*
***Material and Method:*** *A 14-month-old baby was transferred to Siriraj Hospital with the suspicion of basal encephalocele after the surgical repair of cleft lip and cleft palate had been performed.*
***Results:*** *Computerized tomography and magnetic resonance imaging of the head revealed a defect at the anterior cranial fossa and sella floor with a protruded soft tissue mass. The complete radiological examination concluded the diagnosis of TSE. Ophthalmic examination revealed posterior staphyloma and right optic nerve hypoplasia. Transcranial exploration of the anterior skull base showed the protrusion of pituitary stalk and its content into the bony defect at sella turcica, lateral attachment of the olfactory bulb, separated optic chiasm and bilateral optic nerve hypoplasia.*
***Conclusion:*** *Basal encephalocele should be cautious when a soft tissue mass is found within the nasal cavity or at the roof of the mouth in patients with craniofacial dysraphism.*

***Keywords:*** *Trans-sphenoidal encephalocele, Craniofacial dyrhaphism, Craniopharyngeal canal*

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Cephalocele is defined as a protrusion of cranial contents beyond the normal confines of the skull that includes meningocele, encephalomeningocele and hydro-encephalomeningoceles. Nevertheless, the term of "encephaloceles" is frequently used in neurosurgical literature. Encephaloceles are an uncommon entity, which occur in approximately one in every 3,000 to 5,000 live births. Suwanwela C and Suwanwela N introduced the widely used classification system, which includes sincipital (frontal), parietal, occipital, and basal encephaloceles[1]. Basal encephaloceles are the rare form of encephalocele with an estimated incidence of one in every 35,000 live births[2,3]. The current classification of basal encephaloceles based on the location of the bone defect and encephalocele sac was introduced by Pollock et al (1968)[4]. Five subtypes have been described: trans-sphenoidal (sphenopharyngeal), sphenomaxill-ary, sphenoethmoidal, spheno-orbital, and transethmoidal. Trans-sphenoidal encephalocele (TSE) represents the rarest form of basal encephalocele. The incidence of TSE is estimated to be 1/700,000 live births[2]. Sometimes the term of "trans-sphenoidal" and "sphenoethmoidal" subtype was problematic in the literatures which has difference in the extension of the bony defect whether confine only in the sphenoid bone or extension anteriorly to the ethmoid bone[3,4].

This paper is a rare case of TSE with a presentation of craniofacial dysrhaphism.

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## Case Report

A 14-month-old Thai baby was born with a cleft lip and palate. Plastic surgery of the median cleft lip and cleft palate was performed uneventfully at another hospital. During the course of the operation, a soft mass was noticed projecting into the nasal cavity

at the site of the nasal septum. The cleft lip was repaired. Marsupialization of the mass revealed clear fluid, and he was referred to our hospital with the suspicion of cerebrospinal fluid leakage due to basal encephalocele.

The head circumference was 44.5 centimeter. The anterior fontanel was 1x1 centimeter in size. The inter-orbital distance was 2.7 centimeter indicative of hypertelorism.

Eye examination demonstrated wide epicanthus, pseudo-strabismus and roving nystagmus that indicated poor fixation. The anterior segments were normal. The mass was found at the median portion of nasal cavity and roof of the mouth (Fig. 1), but no bulging of the mass into the oral cavity was observed.

The anteroposterior projection of a plain skull film and coronal CT of the head showed an oval-shaped density, 1x2 cm in size, extending down into the nasal cavity (Fig. 2A and 2B). On the lateral view of plain skull film and sagittal CT of the head, the bone defect at the sella turcica and ethmoid sinus could be clearly observed (Fig. 2C and 2D). A three-dimensional computerized tomography (3D-CT) reconstruction of the skull revealed midline bifid maxilla, hypertelorism and elongated optic canal (Fig. 3A). Fundoscopic examination revealed posterior staphyloma and right optic nerve hypoplasia. The 3D-CT revealed a midline bone defect at the pituitary fossa and posterior ethmoid sinus regions and showed a gap filled by an extensive meningocele that extended to the nasopharynx (Fig. 3B). Magnetic resonance imaging (MRI) confirmed

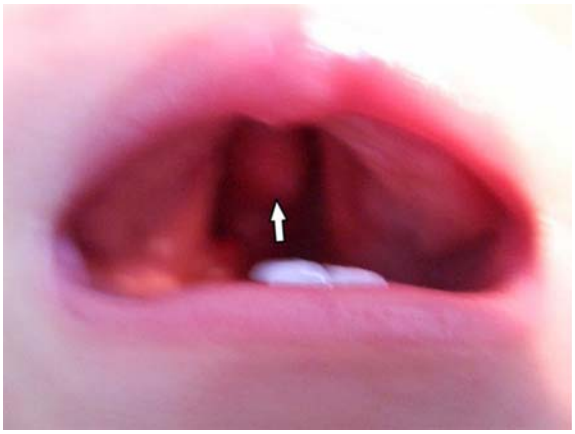

**Fig. 1** Encephalocele at the roof of the mouth (arrow).

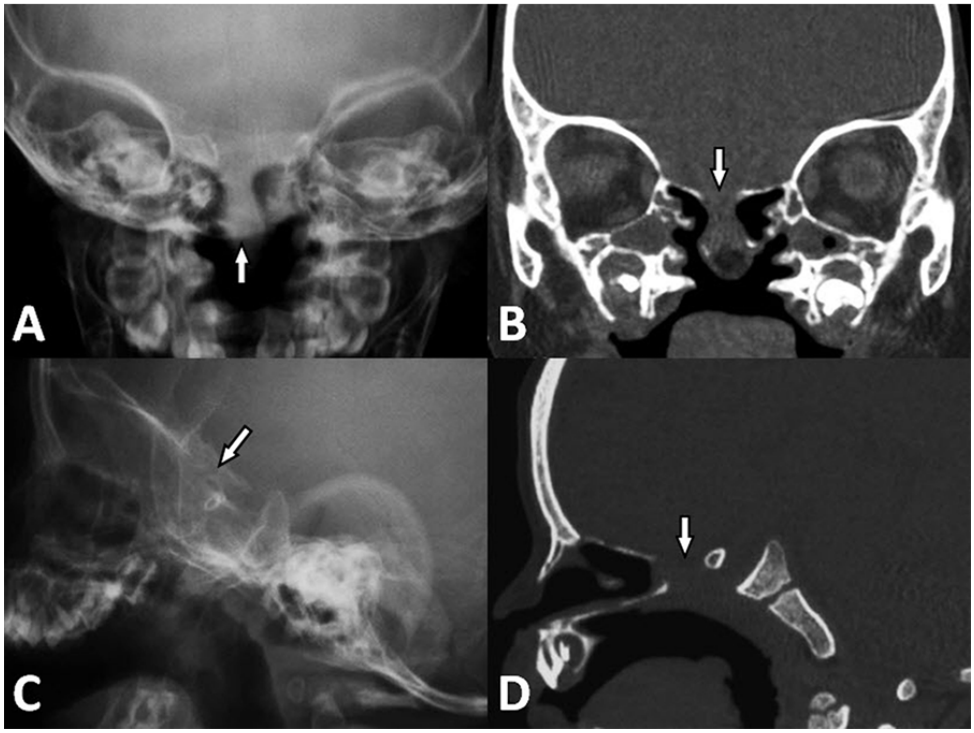


**Fig. 2** A) a frontal skull radiograph shows a soft tissue mass protrudes through the midline defect (arrow); B) a coronal CT of the head shows the defect in the body of the sphenoid bone (arrow); C) a lateral skull radiograph and D) a sagittal CT of the head show the defect at the body of the sphenoid bone (arrow).

the presence of a TSE that produced a soft tissue mass within the posterior nasopharynx and communicating with the third ventricle (Fig. 4A). Other findings included distortion of the intracranial optic nerves, no decussation of optic chiasm, corpus callosum dysgenesis (Fig. 4B), ectopic pituitary gland at inferior margin of the encephalocele, and hypertelorism.

Because of CSF rhinorrhea after surgical repair of cleft palate, we decided to repair the defect transcranially. The patient was in the supine position. A bicoronal incision was used to expose the anterior cavarial and frontonasal regions. A bifrontal craniotomy was made and intracranial exploration was encountered. Intra-operatively, the bone defect was noted at the area of sella turcica with extension of the pituitary stalk, including anterior third ventricle and pituitary gland into the bone defect. Bilateral olfactory nerves were deviated laterally from midline, and bilateral optic nerve

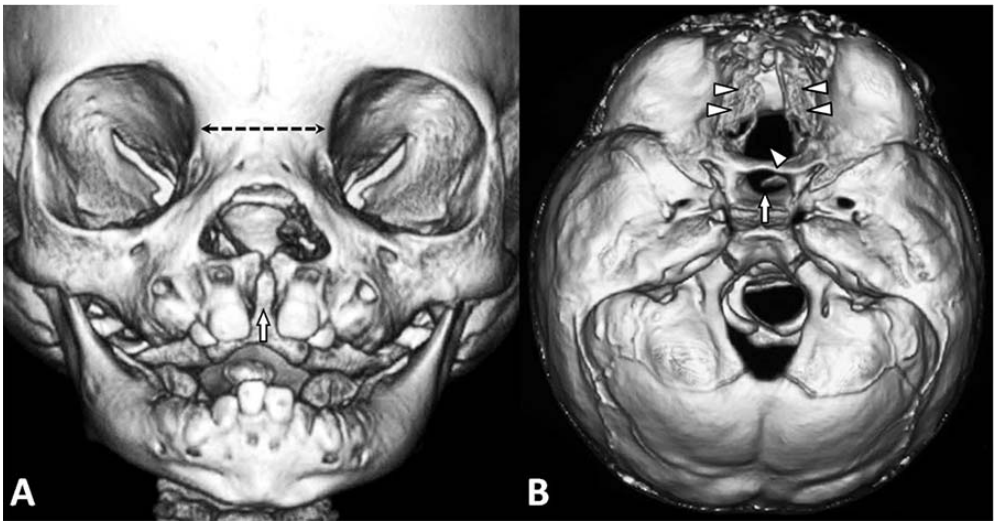


**Fig. 3** A) a 3D-CT reconstruction of the skull reveals midline bifid maxilla (arrow), hypertelorism (dotted bidirectional arrows) and elongated optic canal; B) a 3D-CT reconstruction of the skull base demonstrates basal skull defects at the sella turcica (arrow) and anterior cranial fossa (arrowhead) with lateral displacement of the cribiform plates (double arrowheads).

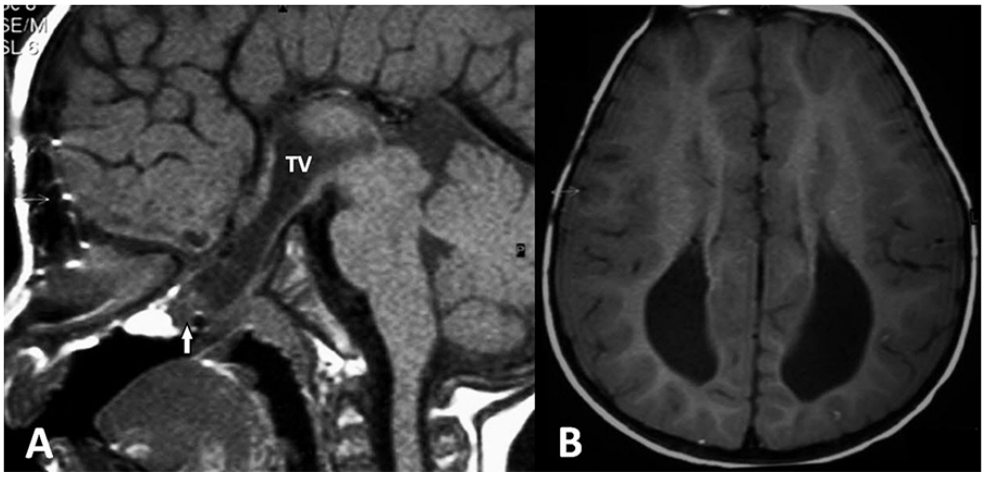


**Fig. 4** A) a coronal T1-weighted MR image demonstrate herniation of third ventricle (TV) through a defect in the pituitary fossa that produced a soft tissue mass within the posterior nasopharynx (arrow) and communicating with the third ventricle (TV); B) an axial T1-weighted MRI shows agenesis of the corpus callosum.

dysplasia extending into optic canal with divided chiasm was also seen (Fig. 5). The protruded content was dissected from the bony defect, dural defect was repaired as watertight fashion and bony defect was closed using split cavarial graft. Because of hypertelorism, medial orbital advancement was performed. There was no cerebrospinal fluid leakage or other postoperative complications.

Due to the critical position of the bony defect, visual and endocrinological abnormalities are frequently associated with basal encephaloceles[5-7]. Extensive endocrine screening for hypothalamic-pituitary dysfunction showed no abnormalities.

## Discussion

### *Pathophysiological mechanism*

Trans-sphenoidal encephalocele (TSE) is a rare type of encephaloceles[1-3,8]. Their cause is unknown but there are many theories explain the development of classic TSE in which the brain including its linings herniate through a defect in the base of the sphenoid bone. The accepted theory explains that there is a developmental defect of ossification of the body of the sphenoid bone as well as persistence of the craniopharyngeal canal (CPC), which normally closes by 50$^{th}$ day of gestation[2,8,9]. The hypothalamus and optic chiasm, together with the third ventricle, usually herniate inferiorly through a defect at the sphenoid bone into the epipharynx because of persistence of CPC. This can cause nasal mass.

### *Clinical presentation and diagnosis*

Koral et al reviewed the literature, of the 43 cases suffered from trans-sphenoidal encephaloceles. Twenty-four out of 43 (57%) had abnormal hypothalamic-pituitary function, 29 (67%) had facial abnormalities with median cleft lip and palate and hypertelorism. Twenty-six (60%) had visual disturbances especially optic atrophy and visual field defects. Eighteen (42%) had central nervous system (CNS) abnormalities especially associated with agenesis of the corpus callosum[4]. In term of sphenoethmoidal encephaloceles, they also found 21 cases. Seventeen of 21 (81%) had midline facial abnormalities, 15 (71%) had visual disturbances, 10 (48%) had pituitary dysfunction, and 12 (57%) had agenesis of the corpus callosum.

Apart from optic atrophy and visual field defect, the presence of morning glory syndrome, coloboma and megalopapilla have been described to be associated with basal encephaloceles[10-12]. However, our case showed only optic nerve hypoplasia.

Several patients have been reported with hypothalamic-pituitary dysfunction but with different pattern of involvement. Growth hormone deficiency, hypothyroidism, central hypogonadism, central adenocortical insufficiency, vasopressin deficiency, and diabetes insipidus have been documented[5-7].

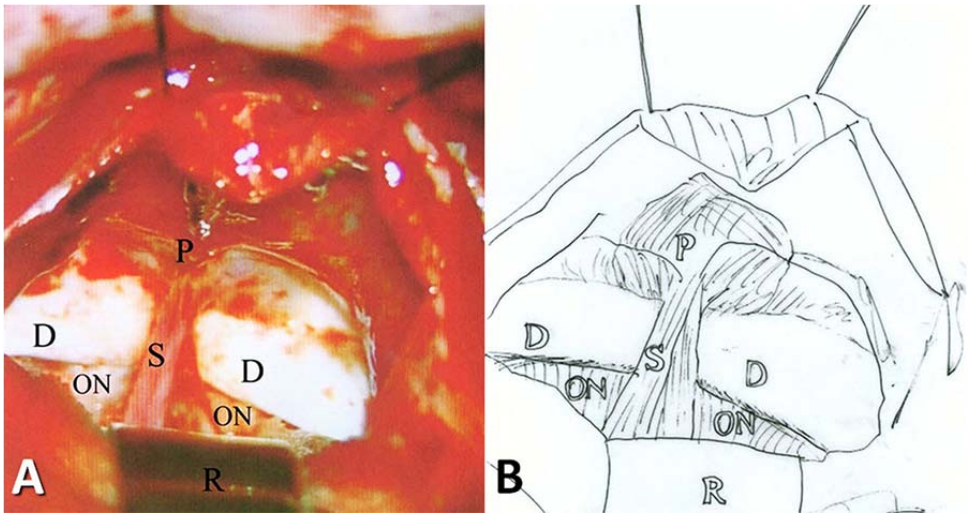


**Fig. 5** A) An intraoperative photograph in a view from the top of the head, the frontal lobes are retracted posteriorly, showing a protrusion of the pituitary stalk (S), anterior portion of third ventricle and pituitary gland (P) into the bony defect in pituitary fossa. The bilateral optic nerves (ON) are shown as fan shape extending to the optic canal; B) a drawing picture of the operative view; D, dura mater; R, retractor.

Agenesis of the corpus callosum is the most cerebral abnormality in TSE suggests this is midline complex anomaly syndrome[13].

### *Surgical treatment of trans-sphenoidal encephaloceles*

To date, the surgical results for TSE have not always been beneficial that may be due to the incomplete assessment of the contents of the encephalocele. Pre-operative evaluation of basal encephaloceles using MR imaging is essential to confirm the extent of the lesion and associated abnormalities and to plan the safest possible repair[14,15]. MR imaging provides the most accurate road map for these delicate interventions. The treatment, indications, and surgical approaches for TSE remain controversial[16-18]. Intervention is indicated for respiratory obstruction, for rhinorrhea or meningitis, and for progressive visual defects due to the compressive lesion[14]. Although TSE have been treated by the transcranial or the transpalatal or the trans-sphenoidal route, the optimal mode of treatment has not yet been established[16-18]. The transcranial approach is associated with high postoperative rates of morbidity, mortality, and hypothalamic dysfunction but this approach has wide exposure to identify all of structure associated with these anomalies. In the transpalatal approach, the closure of the mucosal layer and the reconstruction of the skull base are not simple. The trans-sphenoidal repair seems to be the most suitable approach for TSE in an adult who presented with spontaneous CSF rhinorrhea but this route is not practical in small children.

## Conclusion

Trans-sphenoidal cephalocele (TSE) is a rare condition that associates with other craniofacial, ophthalmic, and brain abnormalities. The basal encephalocele should be considered when there is a soft tissue mass at the area of nasal cavity in the patient with craniofacial dysrhaphism. Complete ophthalmologic, endocrinological and radiological evaluation should be done before the operation. Transpalatal repair in patients with cleft palate is preferred when CSF leakage or nasal obstruction are present. Anyway, transcranial repair can demonstrate these abnormalities.

## What is already known from this topic?

Basal encephalocele, a rare entity of congenital craniofacial malformation, is often associated with aberrant hypothalamic-pituitary function, facial anomalies, dysplasia of the optic apparatus and agenesis of the corpus callosum. Complete pre-operative investigation is beneficial for operative planning.

## What this study adds?

Even though basal encephalocele is relatively rare, it should be highly suspected and investigated in patients with presence of a soft tissue mass in the palate. Surgical treatment is mandatory in symptomatic patients, especially in those who present with spontaneous or iatrogenic CSF leaks.

## Potential conflicts of interest

None.

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## *ภาวะสมองยื่นผ่านรูเปิดของกระดูกสฟีนอยด์ร่วมกับความผิดปกติของการปิดกะโหลกศีรษะและใบหน้า: รายงานผู้ป่วย 1 ราย*

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***ภูมิหลัง:*** *ภาวะสมองยื่นผ่านรูเปิดของกระดูกสฟีนอยด์เป็นภาวะสมองยื่นผ่านฐานกะโหลกศีรษะออกมาทางกระดูกสฟีนอยด์ซึ่งพบได้น้อยมาก*

***วัตถุประสงค์:*** *เพื่อรายงานอาการทางคลินิก ลักษณะภาพทางรังสีและการรักษาโดยการผ่าตัดในผู้ป่วยหนึ่งรายที่มีภาวะสมองยื่นผ่านรูเปิดของกระดูกสฟีนอยด์*

***วัสดุและวิธีการ:*** *เด็กชายอายุ 14 เดือนถูกส่งตัวมารักษาที่โรงพยาบาลศิริราชเนื่องจากสงสัยว่ามีสมองยื่นผ่านฐานกะโหลกศีรษะในระหว่างการผ่าตัดรักษาภาวะปากแหว่งและเพดานโหว่ที่โรงพยาบาลแห่งหนึ่ง*

***ผลการศึกษา:*** *การตรวจเอกซเรย์คอมพิวเตอร์สมองพบมีช่องโหว่ของฐานกะโหลกศีรษะด้านหน้าและบริเวณแอ่งของต่อมใต้สมองร่วมกับมีเงาเนื้อเยื่อผ่านช่องโหว่ดังกล่าว การตรวจภาพแม่เหล็กไฟฟ้าสมองพบว่ามีสมองยื่นผ่านช่องโหว่ดังกล่าว จากการตรวจภาพทางรังสีสามารถให้การวินิจฉัยภาวะสมองยื่นผ่านรูเปิดของกระดูกสฟีนอยด์ การตรวจทางจักษุวิทยาพบการฝ่อของเส้นประสาทตาขวาและลูกตายื่นไปข้างหลังมากกว่าปกติจนไม่เป็นทรงกลม ผู้ป่วยได้รับการผ่าตัดเปิดกะโหลกศีรษะด้านหน้าและตรวจหารูโหว่ของฐานกะโหลกศีรษะเพื่อทำการปิดโดยใช้วิธีการผ่าตัด ทางจุลศัลยกรรมพบความผิดปกติร่วมคือตำแหน่งเส้นประสาทรับกลิ่นอยู่ในตำแหน่งด้านข้างมากกว่าปกติ optic chiasm แยกออกจากกัน และมีการฝ่อของเส้นประสาทตา*

***สรุป:*** *ควรระมัดระวังภาวะสมองยื่นผ่านฐานกะโหลกศีรษะในกรณีที่พบก้อนในโพรงจมูกหรือที่เพดานปากในผู้ป่วยที่มีความผิดปกติของการปิดกะโหลกศีรษะและใบหน้า*