

## Case Report

# A Rare Case Report of Total Aplasia of Paranasal Air Sinuses with Posterior Fossa Malformation (Dandy Walker)

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## Abstract

**Background:** Total aplasia of paranasal air sinuses is a very rare condition. Dandy Walker malformation is the most common posterior fossa malformation involving cerebellum and IV ventricle. We present herewith a rare case of aplasia of paranasal sinuses with Dandy Walker malformation. There is no parallel case reports of these combined anomalies found in one patient available to our knowledge.

**Case report:** A mentally retarded 7 years old male child was referred for CT scan study of P.N.S. and Brain. The C.T. findings of P.N.S. and Brain showed aplasia of all paranasal air sinuses and characteristic of Dandy Walker malformation such as Hypoplasia of cerebellar vermis, Dilated IV ventricle, Mega cistern magna etc.

**Conclusions:** Total aplasia of paranasal sinuses is a very rare condition. Computed Tomography (CT) scan remains the gold standard in revealing anatomic details of paranasal sinuses. Dandy-Walker malformation is a rare congenital anomaly of posterior fossa involving cerebellum and IV<sup>th</sup> ventricle. Treatment depends upon the severity of abnormality and status of patient at the time of its detection.

## Key words

Aplasia, Cerebellum, Malformation, Paranasal, Posterior fossa, Sinus.

## **Introduction**

The paranasal air sinuses are air-containing cavities in the bones around the nasal cavity. The development of paranasal sinuses and their final shapes exhibit great variations, even between the twins. Fractures, tumors, mucoceles, primary ciliary dyskinesia, infections, and some syndromes such as Down's syndrome may have adverse effects on paranasal sinus development.

The hypoplasia or aplasia of paranasal air sinuses is a rare phenomenon that mainly affects the frontal (12%) and secondarily to the maxillary sinuses (5 to 6%). Aplasia of sphenoidal sinuses is extremely rare. Simultaneous aplasia of multiple sinuses is a still rarer finding.

Computed tomography (CT) Scan is an excellent imaging modality to document the detailed anatomy of the paranasal sinuses.

Dandy-Walker syndrome is a rare congenital anomaly involving cerebellum and IV<sup>th</sup> ventricle. The condition is characterized by aplasia or hypoplasia of the cerebellar vermis, cystic dilatation of IV<sup>th</sup> ventricle and enlargement of posterior fossa. The estimated prevalence of a Dandy-walker malformation is about 1 per 30,000 live births. Clinical presentation depends on the severity of the abnormality.

We present here a case report of total aplasia of paranasal air sinuses associated with Dandy-Walker malformation – combination of rare of the rarest case.

## **Case report**

A male child aged 07 years presented to ENT surgeon with repeated upper respiratory tract infections and thereafter referred for computed tomography study of P.N.S. and Brain.

Patient was borderline mentally retarded. No associated neurological symptoms or cerebellar signs were noted. No other craniofacial anomaly was observed. He had no previous history of facial trauma, irradiation or systemic diseases

affecting the skeletal system such as Paget's disease, osteopetrosis, or fibrous dysplasia.

The patients axial and coronal CT scans (2 mm slice thickness) of paranasal sinuses and brain were done.

## **Observations**

The C.T. findings of P.N.S. and Brain were as follows -

- Aplasia of entire paranasal air sinuses viz aplasia of frontal, sphenoidal, maxillary and ethmoidal air sinuses without pneumatization on either side.
- Normal nasal septum and turbinates.
- Craniostenosis (deformity of skull due to premature closure of cranial sutures)
- Dilated IV<sup>th</sup> ventricle adjacent to its communication with cisterna magna.
- Mega cisterna magna communicating with IV<sup>th</sup> ventricle and lateral C.S.F. spaces.
- Hypoplasia of cerebellar vermis.
- Mild asymmetrical dilation of left lateral ventricle.
- Fullness of III<sup>rd</sup> ventricle.
- All these findings were suggestive of dandy-walker malformation associated with aplasia of paranasal air sinuses.

**Images 1 to 6** indicate total aplasia of paranasal sinuses and premature closure of cranial sutures (Craniostenosis) at 07 year. **Images 7 to 12** fulfill triad of Dandy Walker Malformation.

## **Discussion**

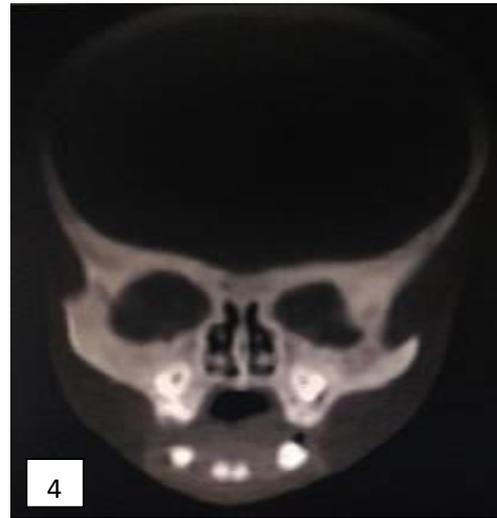
Although the development of paranasal sinuses begins in the 3<sup>rd</sup> week of gestation, their expansion continues after birth throughout early adulthood alongwith the development of facial cranium and teeth [1]. They develop as mucosal diverticula of the main nasal cavity invading the adjacent bones.

Maxillary and ethmoidal sinuses are presented at birth as rudimentary air cells, and sphenoidal and frontal sinuses are not present at birth.

**Image – 1:** Aplasia of frontal sinuses (no pneumatization).



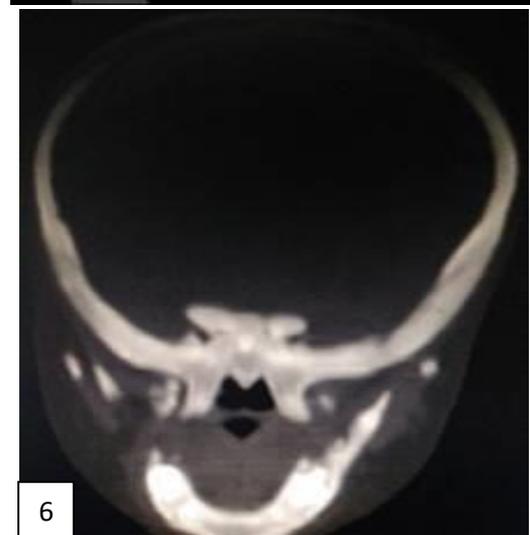
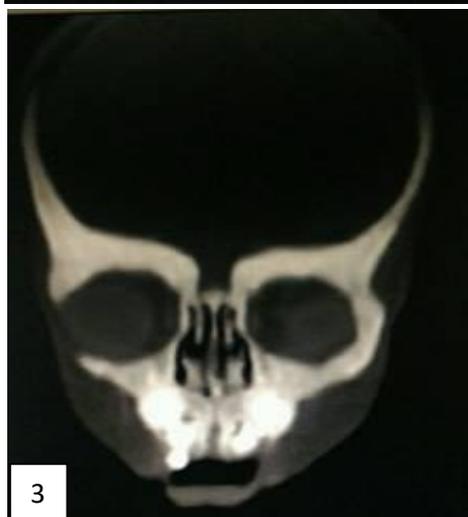
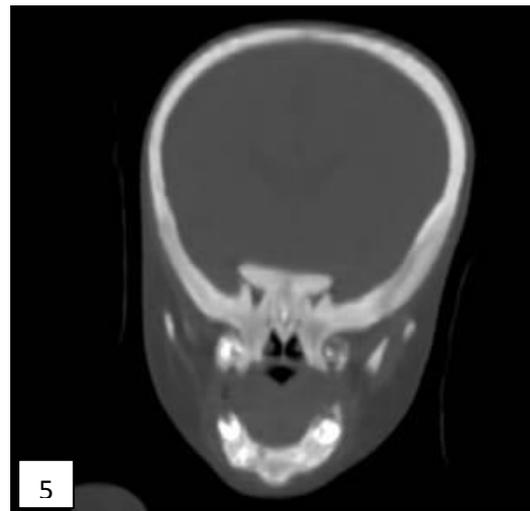
**Image – 4:** Maxillary sinus aplasia. There was normal vault thickening and no bony asymmetry excluding Fibrous dysplasia.



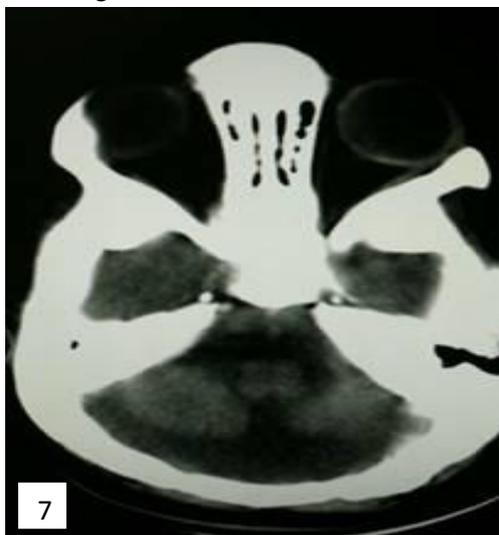
**Image - 2, 3, 4:** Aplasia of ethmoidal sinuses. Note middle turbinate is arising from cephalad portion of nasal cavity indicating that ethmoidal sinuses have not developed. Nasal septum and turbinates have developed normally.



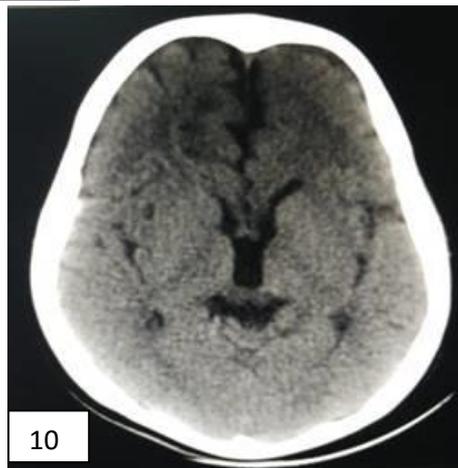
**Image – 5, 6:** Aplasia of Sphenoidal sinuses. (Normal development starts at 2-3 years after birth).



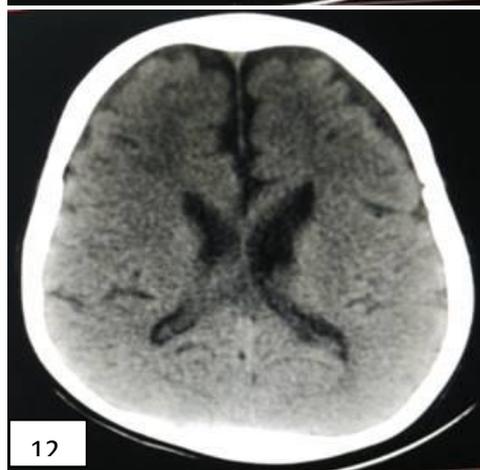
**Image – 7, 8, 9:** Mega cisterna magna communicating with IV<sup>th</sup> ventricle and Hypoplasia of cerebellar vermis, Dilated IV<sup>th</sup> ventricle adjacent to its communication with cisterna magna.



**Image - 10:** Fullness of III<sup>rd</sup> ventricle.



**Image – 11, 12:** Mild asymmetrical dilation of left lateral ventricle.



Maxillary sinus starts developing during the intrauterine third month. Lower boundaries of the sinus and the nasal cavity reach the same plane around the age of 10 years. This development continues until puberty [2]. The maxillary sinus extends laterally past the infraorbital canal by the

age of 4 years and reaches the maxillary bone by the age of 9 years. At birth, ethmoid cells are more developed anteriorly and pneumatization progresses in a posterior direction. Their growth lasts until late puberty. Pneumatization of the sphenoidal sinuses can be detected as early as 2-3 years of age and they reach the final size by the age of 12-14 years. Sphenoidal sinus is located in the sphenoid bone and its shape and size varies between individuals. Sphenoidal sinus aplasia is rare and has been reported to be 1-1.5% and may present with craniofacial abnormalities and syndromes. The last paranasal sinus undergoing major expansion is the frontal sinus. Its growth starts 2 years after birth and reaches its final size after puberty.

The aplasia of the paranasal sinuses occurs more frequently in syndromes of craniosynostosis, osteodysplasia (Melnick-Needles), as well as in cases of Down's syndrome (hypoplasia of the frontal sinus) [3]. The prevalence of frontal and sphenoidal sinus aplasia or hypoplasia have been shown to be higher in patients with cystic fibrosis or primary ciliary dyskinesia.

Paranasal sinuses present great structural variations.

Frontal sinus is located posterior to the superciliary arches between the internal and external portions of the frontal bone. Ventilation of this sinus varies individually, and it is also different for the right and left halves. Frontal sinus develops very slowly. Ventilation of the sinus starts around the age of two years, and its development continues until adolescence.

Frontal sinus aplasia is present unilaterally in 15% and bilaterally in 5% of normal adults [4].

However, frequency of bilateral absence of the frontal sinuses was significantly higher in some populations, including Alaskan Eskimos (25% in males and 36% in females) and Canadian Eskimos (43% in males and 40% in females) [5, 6].

Functional roles of paranasal sinuses continue to be elusive [7]. Some of the functions ascribed to these sinuses are as follows:

- They lighten the skull.
- Impart resonance to the voice.
- Serve as air conditioning chambers by adding humidity and temperature to the inspired air.
- Assist in facial growth and architecture.
- Function as pillars for dispersal of masticatory forces.
- Provide protection for the brain.
- Provide thermal insulation for the central nervous system and sense organs.
- Serve to increase surface area of the olfactory mucosa.
- Provide even distribution of inspired air, which aids in olfaction.

The variations in the anatomy of the frontal sinus may be critical for morphological or forensic investigations and for neurosurgeons performing pterional or supraorbital craniotomy because of the proximity of the sinus to the orbit and the anterior skull base [8].

Many patients with maxillary sinus aplasia are asymptomatic, and the condition is incidentally detected on routine radiography. However, some patients present with chronic headaches, facial aches, and voice problems. Unilateral opacified appearance of the maxillary sinus on plain radiography may be seen as a result of infectious mucosal thickening, tumoral involvement due to chronic sinusitis or aplasia/hypoplasia [9].

Some reasons have been mentioned as a cause of congenital maxillary hypoplasia or aplasia, such as:

- Arresting of the development because of infection, injuries, and irradiation.
- Congenital first arch syndrome.
- Developmental anomalies such as craniosynostosis,

osteodysplasia, and Down syndrome.

Also, some reasons are responsible for an acquired category of maxillary sinus hypoplasia, such as:

- Trauma with deformity due to fracture or surgery in the sinus region.
- Thalassemia and cretinism.
- Wegener's granuloma (inflammatory osteitis).
- Neoplasms that cause osteitis.

Maxillary sinus aplasia is extremely rare, whereas maxillary sinus hypoplasia is a well-known clinical entity [10]. Maxillary sinus hypoplasia is reported in 1.7–10.4% of patients, while its aplasia has been reported to be 5–6%.

Recognition of maxillary sinus aplasia is particularly important in preventing damage to the orbit during endoscopic sinus surgery. Aplasia of the sphenoid sinus is extremely rare; however, recognition of this condition in advance is crucial in patients who are to undergo trans-sphenoidal hypophysectomy.

In addition, clinical presentations and complaints of patients with PNS aplasia can provide information about the functions of PNSs. In conclusion, aplasia of PNSs can point to clinically significant diseases and it must be kept in mind and explored during planning for surgical intervention to the sinuses in order to prevent complications.

Headache, facial pain, nasal drip, and voice problems can be observed in patients with PNS aplasia but most patients are usually asymptomatic or not aware of the symptoms. Paranasal sinus aplasia decreases the threshold of pain occurrence. Patient is devoid of the functions of the paranasal sinuses such as skull lightening, thermal insulation for central nervous system, and sense organs. Therefore, any irritating factor such as cold air, chemical

irritants, stress, and anxiety easily triggers the headache. Patient may experience masseter tiredness on chewing, which points out another function of the paranasal sinuses, which is dispersal of masticatory forces on chewing.

Simultaneous aplasia of multiple sinuses is a rare event. Khanduri, et al. [11] reported a 54-year-old female patient with the complaints of nasal obstruction and persistent headaches, who was found to have bilateral frontal and sphenoid sinus aplasia along with bilateral maxillary and ethmoid sinus hypoplasia. Korkmaz, et al. [12] reported a 57-year-old female patient with complaints of nasal fullness and chronic episodes of headaches, who had total aplasia in all paranasal sinuses.

Dandy-Walker malformation (DWM) was initially described by Walter Dandy and Arthur Earl Walker in 1914. It is the most common posterior fossa malformation, characterised by the triad of:

- Hypoplasia of the vermis and cephalad rotation of the vermian remnant
- Cystic dilatation of the fourth ventricle extending posteriorly, usually the cerebellar hemispheres are displaced anterolaterally, but with a normal size and morphology.
- Enlarged posterior fossa with torcula-lambdoid inversion (torcula lying above the level of the lambdoid due to abnormally high tentorium)

### **Epidemiology**

The estimated prevalence of a Dandy-Walker malformation is about 1 per 30,000 live births and accounts for 4–12% of the cases of infantile hydrocephalus.

### **Clinical presentation**

The syndrome can appear dramatically or develop unnoticed. Clinical presentation is dependent on the severity of the abnormality. In a classic Dandy-Walker malformation, patients usually manifest in the first year of life with

symptoms of hydrocephalus and associated neurological symptoms. Macrocephaly is the most common manifestation and in 80% of cases, the diagnosis is made by the first year of life. Despite severe cerebellar abnormalities, cerebellar signs are not common.

Up to half of affected individuals have intellectual disability that ranges from mild to severe, and those with normal intelligence may have learning disabilities. Children with Dandy-Walker malformation often have delayed development, particularly a delay in motor skills such as crawling, walking and coordinating movements. People with Dandy-Walker malformation may experience muscle stiffness and partial paralysis of the lower limbs (spastic paraplegia), and they may also have seizures.

In older children, symptoms of increased intracranial pressure such as irritability, vomiting, and convulsions and signs of cerebellar dysfunction such as unsteadiness and lack of muscle coordination or jerky movements of the eyes may occur.

### Differential diagnosis

The differential is that of other causes of an enlarged CSF retrocerebellar space (and mimics thereof) including:

- Mega cisterna magna
- Epidermoid cyst
- Arachnoid cyst
- Blake pouch cyst

Dandy-Walker malformation may be isolated or can occur with other malformations of the brain.

Associated CNS abnormalities include -

- Aplasia or dysgenesis of the corpus callosum
- Lipomas of the corpus callosum
- Aqueductal stenosis
- Microcephaly
- Encephalocele.
- Lumbosacral meningocele
- Cleft palate
- Facial angiomas

- Low-set ears
- Polydactyly or syndactyly
- Cardiac anomalies

Approximately 70-90% of patients have hydrocephalus, which often develops postnatally. Dandy-Walker malformation may be associated with atresia of the foramen of Magendie and, possibly, the foramen of Luschka. Etiology is from hindbrain development arrest. No IV<sup>th</sup> ventricle outlet foramina are formed.

### Treatment

- Treatment may involve physical therapy, special education, or surgical placement of a cerebral shunt.

### Conclusions

Total aplasia of paranasal sinuses is a very rare condition.

Computed Tomography (CT) scan remains the gold standard in revealing anatomic details of paranasal sinuses and to delineate the associated structural abnormalities and pathologies of nasal fossa, which have utmost importance in endoscopic treatment planning.

Dandy-Walker malformation is a rare congenital anomaly of posterior fossa involving cerebellum and IV<sup>th</sup> ventricle. Treatment depends upon the severity of abnormality and status of patient at the time of its detection.

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