Craniosynostosis, Dandy-Walker Variant (DWv) and Arnold-Chiari Malformations — Signs and Symptoms

See online here
These are a group of congenital malformations that involve the cranial cavity and the central nervous system. The Dandy-Walker variant is the commonest posterior fossa malformation, while craniosynostosis is the premature fusion of one or more of the cranial sutures causing abnormal shape of the head. They present with hydrocephalus and features of increased intracranial pressure, thus, the goal of therapy is to relieve the pressure. The main modalities of treatment are surgical interventions either endoscopically or under direct vision via scalp incisions.

Definition of Craniosynostosis, DWv, and Arnold-Chiari Malformations
These are a group of congenital malformations that involve the cranial cavity and the central nervous system.
Dandy walker malformations

Dandy walker malformations are a group of congenital brain abnormalities involving the cerebellum leading to dilatation of the 4th ventricle, poor flow of cerebrospinal fluid and ultimately obstruction of the flow. The dandy walker variant is the less severe form of the spectrum as it doesn’t meet all the criteria to be classified as a malformation.

Arnold-Chiari malformations

Arnold-Chiari malformations are a group of congenital abnormalities that are associated with a bony base of the cranium causing limitation of space in the posterior fossa thus affects the cerebellum, brainstem, and upper spinal cord.

Craniosynostosis

Craniosynostosis is the premature fusion of one or more of the cranial sutures causing abnormal shape of the head.

Epidemiology of Craniosynostosis, DWv, and Arnold-Chiari Malformations

The dandy walker variant is seen in 1 in 30000 people with a sporadic distribution.

They are rare deformities and have no sexual or racial predilection except the Arnold-Chiari malformation which is more common in females. The cause of the malformations is mainly environmental influence in a genetically predisposed individual.

Arnold-Chiari malformations are more common in females with a female to male ratio of 1.3:1 and affect 1 in 1000 live births in the general population.

Craniosynostosis affects approximately 0.1 % of Americans with the secondary craniosynostosis being more common. It does not have any sexual or racial predilection and mainly presents with features of raised intracranial pressures.

Etiology of Craniosynostosis, DWv, and Arnold-Chiari Malformations

Dandy-Walker variant
Environmental influence such as infections with TORCH complex organisms causes congenital malformation.

Increased use of teratogenic drugs such as warfarin leads to a distorted architecture of the brain cavity and deformity formation. Idiopathic in a majority of the cases.

Genetic influence is thought to play a role with an autosomal recessive inheritance pattern being demonstrated.

Arnold-Chiari malformation

Genetic influence is thought to play a key role. It is linked to chromosome 9 and 15. Expression of these genes results into a para-axial mesodermal defect that subsequently results into a small posterior fossa formation. This limits the growth of the cerebellum and nearby structures forcing it to look for growth space in the nearby spinal cavity by herniation. Or an upward herniation of the vermis.

Since the disease is associated with myelomeningocele, it has led to an association that the loss of CSF causes an inadequacy of the stimulus for condensation of the skull base and, thus, results into limited space for growth but enough opening for herniation of the contents.

Craniosynostosis

The non-syndromic variety

- Genetic mutations
- Environmental influence
The syndromic variety

Crouzon’s Syndrome

It is the most common and is associated with bilateral coronal craniosynostosis, midfacial abnormalities, hydrocephalus, forward protrusion of the eyes and airway obstruction. In one-third of patients, the syndrome is thought to arise from a mutation localized in the GFR2 gene.

Apert’s Syndrome

This syndrome is also characterized by other forms of synostosis in addition to bilateral craniosynostosis. It is also associated with hand, elbow, hip, and knee deformities with forwarding protrusion of the eyes. It occurs in about one in 100,000 births.

Carpenter’s Syndrome

This syndrome characterized by lambdoid and sagittal synostosis with limb abnormalities that may include extra digits on the feet and abnormalities of the heart. A genetic location for this syndrome has not yet been identified.

Pfeiffer’s Syndrome

This is a syndrome with unicoronal craniosynostosis, facial deformities, limb abnormalities, protruding eyes, and hearing loss. Three types have been described, with types 2 and 3 being the more severe forms.

Pathophysiology of Craniosynostosis, DWv, and Arnold-Chiari Malformation

Normal skull architecture and enlargement

The development of the brain and skull is coordinated to allow for development of a cranial cavity that only allows cranial contents to fit in and once the development is over the cranial bones permanently fuse with each other making it difficult to accommodate foreign components. The development begins from encoding of the genetic material, intrauterine organogenesis, and postnatal growth of the child.

Any disruption during these periods results into one of the three malformations or other malformations. Some common disruptions in the growth include genetic mutations that give wrong instructions for development and influence by teratogenic toxins such as drugs.

After birth, the normal skull is made up of paired and unpaired bones that are in close association to each other via the metopic, coronal, sagittal and lambdoid sutures. The sutures unite at the anterior and posterior fontanelle all of which fuse normally before the age of two years. Any form of fusion before the full development of the brain leads to microcephaly and limited growth and maturation of the brain. If growth continues, there is increase in intracranial pressure that causes headache, papilledema, and other features.
Dandy-Walker variant

Dandy-Walker variant arises from a genetic or intrauterine influence on the cerebellum that renders the 4th ventricle dilated resulting to **poor flow and obstruction of CSF flow** thus may present with **communicating hydrocephalus**. CSF is produced by the choroid plexus of the lateral ventricles via active secretion and diffusion.

From the two lateral ventricles, fluid drains through the foramen of Monro on each side into a single midline third ventricle. The third ventricle connects by the aqueduct of Sylvius to a midline fourth ventricle that has three exit openings, paired lateral foramina of Luschka, and a midline foramen of Magendie.

These openings lead to a system of interconnecting and focally enlarged areas of subarachnoid spaces referred to as cisterns which allow for free flow of CSF. The **4th ventricle is related to the cerebellum and the cerebellar vermis all of which are deformed in DWv, thus, the patients may present with hydrocephalus.**

**Classification of Craniosynostosis, DWv, and Arnold-Chiari Malformation**

**Arnold-Chiari malformations**

Arnold-Chiari malformations are classified according to the degree and types of deformities involved as follows:

**Arnold-Chiari type I**
The commonest form of the spectrum. It is characterized by extension of the lower part of the cerebellum into an opening in the base of the skull.

**Arnold-Chiari type II**

This variety is only seen in spina bifida. The cerebellum and brainstem herniate into the opening in the base of the skull.

**Arnold-Chiari type III**

It is a severe form of the Chiari malformations with severe neurological deficits due to compression of the spinal cord by herniating cerebellum and brainstem.

**Arnold-Chiari type IV**

This is an incomplete or underdeveloped cerebellum associated with exposed skull and spinal cord.

**Craniosynostosis**

Craniosynostosis can be classified into:

**Simple craniosynostosis**

This is a form of the disease where **only one suture is prematurely fused**. The skull takes a certain shape depending on the fused suture such as:

- Fusion of the sagittal suture leads to a **long and narrow head** (scaphocephaly). This gives the skull a boat like shape and frontal bossing. It is the most common form of craniosynostosis.
- Fusion of the coronal suture leads to **flattening of the forehead** (anterior plagiocephaly)
- Fusion of the lambdoid suture leads to **flattening at the back of the skull and the ear** (posterior plagiocephaly)
- Fusion of the metopic suture leads to **triangular shaped head** (trigonocephaly). The forehead is narrow and eyes close together.
Complex/compound craniosynostosis

A form of the disease where there is a **generalized fusion of the cranial sutures**. It is associated with general body deformities such as Alpert syndrome and Carpenter’s syndrome, hence, known as *syndromic craniosynostosis*. Fusion of multiple sutures also gives various characteristic malformations such as:

- Fusion of the coronal suture bilaterally leads to a **skull wider than normal** (anterior brachycephaly)
- Bilambdoid suture fusion causes a **skull that is wider than normal** (posterior brachycephaly)
- Fusion of the sagittal and metopic sutures leads to **scaphocephaly**.
- Fusion of severe sutures (multisuture fusion) leads to development of a **cloverleaf skull** (Klee blatts).

**Dandy walker malformations**

Dandy walker malformations span a wide spectrum of deformities, including:

- Dandy walker syndrome malformation which is the severe form of the disease.
- Dandy walker syndrome cisterna magna
- Dandy walker syndrome variant which is the less severe form that doesn’t meet the criteria for the above diseases.

**Clinical features of Craniosynostosis, DWv, and Arnold-Chiari Malformation**
Dandy-Walker variant

Clinical presentation of the dandy walker variant is usually before one year of age and entails:

- Features of hydrocephalus and macrocephaly.
- Irritability
- Seizures, vomiting, and hotness of body.
- Delayed developmental milestones such as crawling, walking, and coordination.
- Muscle stiffness, paralysis, and paraplegia.
- Auditory and visual problems.

Craniosynostosis

In craniosynostosis, the patients present with the described deformities:

- Microcephaly that suggests secondary craniosynostosis.
- Positional molding especially in posterior plagiocephaly.
- Torticollis.
- Frontal bossing.
- Pointed forehead/ trigonocephaly infusion of metopic suture.

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<tr>
<th>Localized</th>
<th>Generalized</th>
<th>Generalized with symptoms</th>
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<tr>
<td>Sagittal suture</td>
<td>Multiple sutures</td>
<td>Syndactyly with Apert syndrome</td>
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<td>Coronal suture</td>
<td>Microcephaly</td>
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<td>Lambdoid suture</td>
<td>Developmental delay</td>
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Chiari malformations

Chiari malformations are asymptomatic but when severe they manifest clinically as

Arnold-Chiari Type I

- Ataxia, dysmetria, nystagmus and disequilibrium due to compression of the cerebellum.
- Occipital headache worse on Valsalva maneuver due to increased intracranial pressure.
- Neck pain.
- Features of hydrocephalus.
- Syringomyelia and central cord syndrome.
- Sleep apnea.

Arnold-Chiari type II

- Mainly signs of brainstem dysfunction such as
- Swallowing and feeding difficulties.
- Stridor/difficulty in breathing.
- Apnea.
- Weak cry.
- Nystagmus
- Occipital headache.
Investigations of Craniosynostosis, DWv, and Arnold-Chiari Malformation

The diagnostic investigations include:

**Antenatal diagnosis**

- Obstetric ultrasounds: makes the diagnosis of congenital abnormalities such as absent vermis from the 18th week of gestation.
- Amniocentesis and karyotyping.

**Ultrasound of the head:** preferred imaging in neonates as it is free of complications such as radiation exposure and hypothermia during exposure. Identifies dilated ventricles and fused sutures easily.

**MRI scan of the head:** has similar use as the CT but it is more superior in brain tissue assessment. Thus, it is best for assessment of anatomy, congenital malformations, and deformities such as syringomyelia.

**CT scan of the head:** excellent method of imaging the brain in case of associated bone abnormalities. Preferred when MRI is contraindicated.

**X-rays of the skull:** Anteroposterior, lateral, and special views may be done for craniosynostosis assessment.

**Serial measurements of head circumference across the percentiles** gives the diagnosis of macrocephaly or hydrocephalus if the measure exceeds the 98th percentile. Could also give a hint of microcephaly.

**Complete blood count (CBC) and Renal function tests:** May be done in preparation for theatre and as a guide for use of contrast in other imaging modalities.

**Differential diagnosis of Craniosynostosis, DWv, and Arnold-Chiari Malformation**

- **Hydrocephalus**
  - Most of the conditions present with features of hydrocephalus/ are causes of hydrocephalus.

- **Benign Tumors**
  - They are the underlying causes of hydrocephalus and differentiated on imaging characteristics, and location.
  - Dandy walker variant is associated with meningomyelocele but

- **Neural tube defects**
  - can be differentiated on absence of cerebella abnormalities in neuro tube defects.

- **Pediatric torticollis**

**Treatment of Craniosynostosis, DWv, and Arnold-Chiari Malformation**

It is **predominantly surgical** with some medical approaches being instituted in some cases, such as:
Conservative management

1. Analgesics for headache and neck pains.
3. Cervical collar to stabilize patients neck.
4. Physical/occupational therapy is indicated in dandy walker variant.
5. Special education in the event of delayed developmental milestones.

Surgical therapy

In Arnold-Chiari malformation

The aim of surgical intervention in Arnold-Chiari malformation is to decompress the cervico-medullary junction and allow for the cerebrospinal fluid to flow freely. Interventional options include:

- Posterior fossa craniectomy which involves the removal of the irregular bony architecture of the skull to give the cerebellum more space to expand.
- Electrocautery of the cerebellum via high frequency electric currents to induce shrinkage.
- Spinal laminectomy to remove the pathological bony roof of the spinal canal.

In dandy walker variant and craniosynostosis

Surgical intervention is warranted in situations of hydrocephalus and extreme intracranial pressure elevation. The surgical options available include:

- **Shunting** which involves the surgical placement of a one-way valve drainage system from the proximal ventricle to the distal drainage system through the subcutaneous tissue.
- **Ventriculoperitoneal shunting** involves diversion of the excess CSF flow into the peritoneal cavity.
- **Ventriculopleural shunting** involves diversion of the excess CSF into the large vessels of the heart that eventually drain into the right atrium.

Other surgical options for correction of craniosynostosis include:

- Endoscopic correction of the fused sutures in children younger than 6 months
- Open surgical correction under direct vision via a scalp incision
- Grown enough to withstand surgical stress.

Complications of Craniosynostosis, DWv, and Arnold-Chiari Malformation

Complications of craniosynostosis

1. Permanent facial deformity
2. Poor self-esteem and social isolation
3. In untreated situations, increase in intracranial pressure can lead to developmental delay, cognitive impairment, lethargy, seizures, and death
Complications of Arnold-Chiari malformations

1. Pseudo meningocele formation and cerebrospinal fluid leakage
2. Meningitis and wound infection post-surgery
3. Neurovascular injury during surgery causes deficits and hemorrhage
4. Cerebellar ptosis in large occipital craniectomy

Complications associated with shunts

- **Shunt malfunction**: it is estimated that up to 40% of the inserted shunts fail within the first year. The rate reduces drastically to around 5% after one year.
- **Infection**: infections may arise from the skin during the procedure or the overgrowth of normal gut flora.
- **Overdrainage**
- **Underdrainage**: manifests by the persistence of hydrocephalus signs and complications even after placement of the shunt.
- **Subdural hematoma**: Arises from traumatic injury to the vascular system during stent insertion
- **Seizures**
- **Abdominal complications**: excessive fluid flow into the abdomen results into ascites and increase in intra-abdominal pressure which is responsible for hernia development

Course and prognosis of Craniosynostosis, DWv, and Arnold-Chiari Malformation

Chiari malformations have a **favorable prognosis** with normal life expected after intervention, although some deformities have occurred after intervention.

References


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Lee, J. C. (2016). *Craniosynostosis and Rare Craniofacial Clefts*.

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