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Congenital Varicella Syndrome with Aplasia Cutis Congenita and Aqueduct Stenosis: A Rare Combination

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Abstract: ***Congenital varicella syndrome is a rare disorder characterised by skin scars, limb deficits, ocular and CNS manifestation. However congenital varicella presenting as Aplasia cutis with obstructive hydrocephalus is extremely rare in literature. Aplasia cutis congenita is characterised by localised or generalised condition characterised by absence of skin. Hydrocephalus is a pressure dependent enlargement of the cerebral ventricles. Hydrocephalus caused by aqueductal anomaly is extremely rare. I report a case of Congenital varicella syndrome presenting with generalised tonic clonic seizure, with MRI Brain showing aqueductal stenosis with Aplasia cutis congenita on the scalp.***

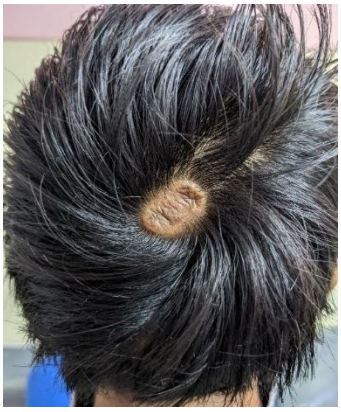
Keywords: Congenital varicella syndrome, Aplasia cutis congenita, hydrocephalus, aqueduct stenosis, vp shunt

1. Introduction

Congenital varicella syndrome refers to spectrum of fetal anomalies associated with maternal varicella zoster infection, generally during first trimester of pregnancy. It is a rare disorder characterised by dermatological manifestations, ocular and CNS manifestation. An uncommon disorder, APLASIA CUTIS CONGENITA, a known complication of congenital varicella, is characterised by absence of skin. A rare CNS manifestation of congenital varicella is Aqueductal Stenosis causing obstructive hydrocephalus. We report a case of congenital varicella syndrome causing APALSIA CUTIS CONGENITA, OBSTRUCTIVE HYDROCEPHALUS which is rare in literature.

2. Case Report

A 15 year old male presented with 2 episodes of generalized tonic clonic seizures for 1 day. It was not associated with fever, decreased sleep, emotional stress, fasting and no history of similar attacks in the past and no significant medical history. There was maternal history of skin lesions characterized by erythematous Papules, macules and crusts, pruritic in nature, at around 14th week of gestation. Her serum varicella zoster specific IgM was positive and she was treated with acyclovir. She was not on any thyroid or anti seizure medications. On physical examination, the patient was alert with normal vital signs, with presence of a localized loss of skin over the scalp, measuring 3*3cm with hair collar sign and no other skin lesions. On CVS examination, the apex beat was found to be on the right midclavicular line and on CNS examination bilateral plantar was extensor. All other physical examinations were unremarkable. The patient was treated with anti seizure medications and then transferred to Neurosurgery department where a right occipital VP shunt was placed. The skin lesion was managed conservatively.



Investigations:

On blood count, TLC-7380/microlitre, PLT-294*10^3/microlitre, Hb-13.5g/dl, RBS-146mg/dl, serum urea-34.7mg/dl, creatinine-0.59mg/dl, serum Na-137mmol/l, normal lipid profile and liver function tests.

MRI brain-Markedly enlarged lateral and third ventricles, suggestive of aqueductal stenosis with obstructive hydrocephalus.

Chest X-ray-normal pulmonary field with heart and it's apex situated on the left side of thorax.

EEG (postictal period)-normal morphology

Varicella zoster specific IgM was negative while serum IgG was 2500 mIU/ml (normal-100mIU/ml).

3. Discussion

The incidence of varicella has been estimated at 0.1–0.7 per 1000 pregnancies, with approximately 2% of fetuses developing congenital varicella syndrome. Congenital varicella syndrome is characterized by a number of clinical manifestations: cutaneous lesions (e.g., scars in a dermatomal distribution and aplasia cutis congenita); CNS and peripheral nervous system abnormalities (e.g., microcephaly, hydrocephalus, cortical/cerebellar atrophy,

mental retardation and intracranial calcifications) and various other systemic manifestations.

Hydrocephalus is a disorder in which an excessive amount of cerebrospinal fluid (CSF) accumulates within the cerebral ventricles and/or subarachnoid spaces, resulting in ventricular dilation and increased intracranial pressure (ICP), it can be communicating or non-communicating type. Aqueductal stenosis is characterized by obstruction of CSF flow through the cerebral aqueduct in the absence of a mass lesion. Aqueductal stenosis is the commonest cause of congenital hydrocephalus. The obstruction of outflow from the third to the fourth ventricle results in triventricular dilation involving the lateral and third ventricles, while sparing the fourth ventricle. Arachnoid webs, strictures, or other glial tissue within the aqueduct are common causes of obstruction. The presenting complaints for hydrocephalus differed according to the age of presentation. Early onset aqueductal stenosis was mainly associated with increase in head circumference, tense anterior fontanelle, sunset eyes. The main complaints for late onset were headache, visual deterioration, urine incontinence, abnormal gait, abnormal behavior and convulsions. Procedures available in the treatment of hydrocephalus include ventriculoperitoneal shunt and more recently Endoscopic Third ventriculostomy. This case was managed with right VP shunt at the neurosurgery department. The Kafarnosky score was estimated preoperative, 1 month post-operative and 6 month post-operative, the score was 54 before operation and improved to 100 after procedure of VP shunt.

KPS score	Description
100	Normal, no complaints, no evidence of disease
90	Able to carry on normal activity; minor signs or symptoms of disease
80	Normal activity with effort, some signs or symptoms of disease
70	Cares for self; unable to carry on normal activity or to do active work
60	Requires occasional assistance, but is able to care for most of his personal needs
50	Requires considerable assistance and frequent medical care
40	Disabled; requires special care and assistance
30	Severely disabled; hospital admission is indicated although death not imminent
20	Very sick; hospital admission necessary: active supportive treatment necessary
10	Moribund; fatal processes progressing rapidly
0	Dead

KPS = Karnofsky Performance Score.

Figure 1: Kafarnosky score of patients aged > 12 years who were diagnosed with hydrocephalus due to aqueductal stenosis. This figure compares the Kafarnosky score pre and post operatively.

Aplasia cutis congenita is characterized by a localized or widespread, complete or partial absence of different layers of the skin at birth. ACC holds an estimated incidence of about 3 in 10, 000 births and there has been a total of approximately 500 reported cases in the literature. The strongest risk factor reported in the literature is the antithyroid drug Methimazole. Frieden classified aplasia cutis congenita into 9 groups. Group 8 refers to aplasia cutis congenita is linked to intrauterine infections (e.g., varicella and herpes simplex).

Pathophysiology of ACC is not well studied and its exact pathogenesis is unknown. However, there are multiple factors that are probably contributing to the development of ACC according to the literature:

1) Chromosomalabnormalities, a recent study has also implicated the UBA2 gene and the SUMOylation pathway
2) Trauma
3) Amniotic irregularities
4) Intrauterine complications, such as vascular accidents or infection
5) Thrombosis, vascular lesions
6) Teratogens: such as misoprostol, benzodiazepines, valproic acid, cocaine, methotrexate, ACE inhibitors, methimazole.

The management of scalp ACC is controversial. Treatment may be either conservative or operative, and there is no consensus or guidelines on treatment strategy. Lesion size is the only universally accepted criterion with larger lesions favoring the surgical approach.

Conservative management consists of regular wound cleansing and application of dressings along with the use of systemic antibiotics. This includes saline drips, betadine solution, bacitracin ointment, and silver sulfadiazine dressings, which are utilized in order to preserve moisture, prevent desiccation, and allow spontaneous epithelialization to occur. Surgical management, on the contrary, includes various procedures. Standard surgical care includes primary wound closure, skin grafting (autologous or allografts), local scalp flaps with or without tissue expansion, free flaps, muscle flaps, full-thickness or split-thickness skin grafts, and cranial vault reconstruction using bone grafts. Specialized surgical techniques such as utilizing bipedicle opposing local flaps, rotational flaps, or L-shaped flaps have been used with satisfying results.

Type	Characteristics
1	ACC located on the scalp with no other anomalies whatsoever
2	ACC located on the scalp but with concomitant limb anomalies such as: Limb malformations (Adams-Oliver syndrome) Hypoplasia or aplasia of the distal phalanges Vascular malformations, fibromas, nipple and hair abnormalities
3	ACC of the scalp along with epidermal nevi, neurological and ophthalmic abnormalities: (such as seizures, mental impairment, corneal and eyelid lesions)
4	ACC accompanied by embryologic deformities: such as omphalocele, leptomeningeal angiomatosis, cranial stenosis, porencephaly, meningomyelocele, spinal dysraphism, or gastroschisis
5	ACC along with fetus papyraceous, placental infarct; Extensive ACC of the trunk or limbs
6	ACC and epidermolysis bullosa involving the lower extremities
7	ACC with no epidermolysis bullosa involving the extremities
8	Teratogen associated ACC: herpes simplex and varicella-zoster virus intrauterine infections, and drugs during pregnancy such as methimazole or carbimazole
9	ACC accompanied by congenital malformations such as: Patau syndrome (Trisomy 13), Wolf-Hirschhorn (4p deletion), Setleis syndrome Johanson-Blizzard syndrome, Goltz syndrome, ADAM complex, Kabuki syndrome Delleman syndrome, Finlay-Mark syndrome, XY gonadal dysgenesis

Figure 2: Frieden classification of ACC types

4. Conclusion

This case of aqueductal stenosis with Aplasia cutis congenita due to congenital varicella syndrome is one of the rarer conditions in medical literature. Aqueductal stenosis responds well to CSF diversion. So a proper maternal and child history is warranted for the diagnosis of congenital varicella syndrome.

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