University Journal of Medicine and Medical Specialities

ISSN 2455- 2852 2018, Vol. 4(4)

A case of Adams-Oliver Syndrome

MANIKANDASAMY V
Department of Neonatology,MADURAI MEDICAL COLLEGE AND HOSPITAL

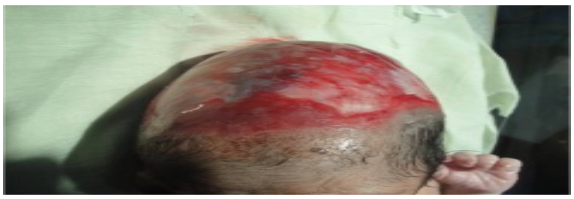
Abstract : A late preterm, 2.3kg, Male child was born to P2L2 mother by Normal Vaginal Delivery. Baby had a large scalp defect, acrania with hypoplastic and absent digits. Acrania portion of the skull was covered by a thick membrane. X-ray skull showed absence of skull vault, X-ray of extremities showed hypoplastic and absent digits. Neurosonogram was normal. Echocardiography done showed moderate PDA with left to right shunt. CT brain was normal. Child was diagnosed as a case of Adams Oliver syndrome. It is an autosomal dominant disorder comprises aplasia cutis congenita with terminal transverse limb defects.

Keyword : Aplasia cutis congenita, Hypoplastic and absent digits

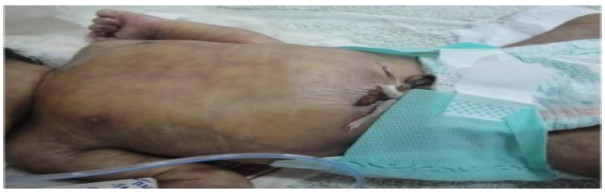
Adams and Oliver described eight members of a family with this disorder in1945. More than 100 affected individuals have been reported.Adams-Oliver syndrome is an autosomal dominant disorder with aplasiacutis congenita and terminal transverse limb defects. It affects both sexesequally.

CASE REPORT

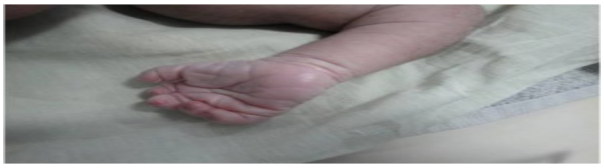
A late preterm, 2.3kg, Male child was born to P2L2 mother by NormalVaginal delivery to a non consanguineously married parents. Baby criedimmediately after birth. Apgar was 5 and 8 at 1 min and 5 min respectively.No adverse perinatal events occurred during birth. Elder sibling was aliveand healthy, had no scalp or limb defects. Both the parents had no scalp orlimb defects. Cry and activity was good. Baby's vitals were within normal limits. Headcircumference is 33cm. Baby had a large scalp defect, acrania withhypoplastic and absent digits. Acrania portion of the skull was covered by athick membrane. Systemic examination findings were within normal limits.

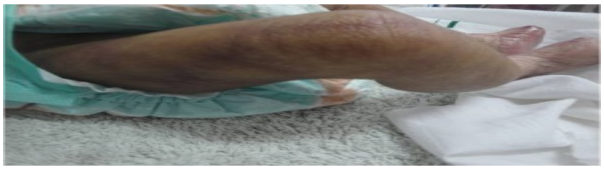


Acrania portion of Scalp



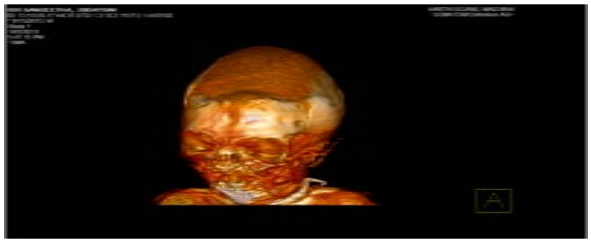
Skin mottling Hypoplastic digits



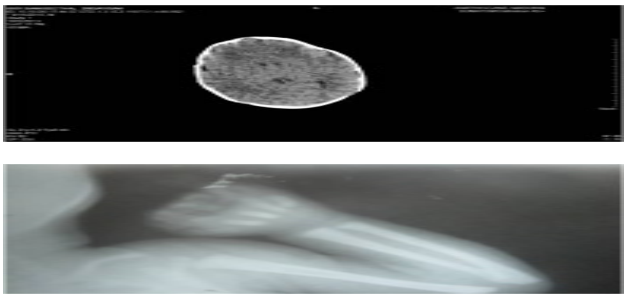


Hypoplastic digits

Baseline investigations were within normal limits. On radiologicalexamination skull showed absence of skull vault, and extremities showedhypoplastic and absent digits. Neurosonogram showed absence of anyintracranial anomalies. CT brain taken showed normal intracranial structures. Baby had moderate PDA with left to right shunt inEchocardiography.



CT brain CT brain



X ray upperlimb

DISCUSSION

In 1945, Forrest Adams and C. Peter Oliver from Minneapolis first reportedthis condition. This condition is transmitted by Autosomal dominant mode oftransmission, with presence of vertex cranial defects resembling aplasiacutis congenita and terminal limb malformations. More than 100 affectedindividuals have been reported.Mild growth deficiency (3rd to 10th percentile) is seen. Aplasia cutiscongenita over posterior parietal region, with or without an underlying defectof bone. Variable degrees of terminal transverse defects, including those oflower legs, feet, hands, fingers, toes, or distal phalanges, short fingers andsmall toe nails. Cardiac defects seen in 20% of affected individuals,including ASD, VSD, Coarctation of Aorta, obstructive lesions of the leftheart, hypoplastic left and right ventricles, DORV, and DOLV. Cutismarmorata telangiectasia seen in 20% of cases.

An autosomal dominant inheritance pattern with marked variability inexpression and lack of penetrance is seen in majority of cases. Gain-of-function mutations of *ARHGAP31, a Cdc42/Rac1 GTPase regulator are*responsible for the defects.Autosomal recessive inheritance has been suggested in a few families.Recessive mutations in the dedicator of cytokinesis 6 (*DOCK6)*gene havebeen identified in some cases. Far more likely to have a severe phenotypewith neurologic abnormalities and intellectual disability.The pathophysiologic mechanism of these defects remains unclear, butseveral mechanisms have been proposed, including predisposition toamniotic rupture sequence, other forms of extrinsic trauma or compression,and vascular compromise. An intrinsic predisposition to interference withnormal tissue development seems a likely etiology. The association of cutismarmorata and the dilated and or tortuous scalp veins may be additionalindicators pointing to an underlying predisposition to vascular compromisein “watershed areas such as the cranial vertex and limbs.

Several syndromes with congenital skin and limb defects have to bedifferentiated from the syndrome of scalp defect with perodactyly . Distaldeficiencies occur in the aglossia-adactylia anomaly (Hanhart syndrome),Poland complex, and as a part of the limb defects in focal dermalhypoplasia. A characteristic pattern of congenital scalp defects can be seenin the syndrome of scalp defects and postaxial polydactyly, the syndrome ofscalp defects and split-hand defect, trisomy 13, and Johanson-Blizzardsyndrome. The skin defects of the amniotic band sequence are rarely foundas localized defects on the scalp. In epidermolysis bullosa dystrophica typeBart the defects are typically on the lower legs, and in focal dermalhypoplasia irregular atrophic areas are observed.Genetic counseling regarding the inheritance of this syndrome should begiven to all parents. In genetic counseling autosomal dominant inheritancewith great variability in expression of the syndrome of congenital scalp defect and distal reduction defects of the limbs should be stressed. However, genetic counseling of families with a sporadicmanifestation of the syndrome may be difficult. An unaffected parent mayrepresent a nonpenetrant individual.

Thus, the unaffected parents of anaffected child always have some risk of having another affected child.Ultrasonic examination might be indicated in all potential affectedpregnancies.Larger scalp defects with underlying defects of bone, where the superiorsagittal sinus or dura are exposed there is an increased risk of hemorrhageor meningitis. Early surgical intervention with grafting is indicated.Cases inwhich the sagittal sinus or dura is not exposed, healing without need forgrafting almost always occurs.Prognosis is excellent in vast majority of cases.

REFERENCES

Whitely CB, Gorlin RJ. Adams Oliver Revisited. American Journal ofMedical genetics 1991: 40: 319-26.

Bamforth JS, Kaurah P, Byrne J, Ferreira P. Adams Oliver Syndrome: Afamily with variability in clinical expression. American Journal of Medicalgenetics 1994: 49: 393-96.

Smith’s recognizable patterns of human malformations 7th edition. P 416.