Acro-Renal Syndrome with Open Lip Schizencephaly – A Rare Case Report

Mojahid Mondal[1], Narayan Pandit[2]

[1, 2] Department of Radio-Diagnosis, North Bengal Medical College and Hospital, Siliguri, West Bengal, India.

PRESENTATION OF CASE

A 7-day-old new-born was referred to Department of Radiodiagnosis of North Bengal Medical College and Hospital to evaluate enlargement of head circumference. On clinical examination of this new-born we found macrocephaly and absent right thumb as well as deformed left thumb. No other clinical abnormalities were seen in this new-born apart from this. Facial structures, spine, upper and lower limbs, trunks were normal clinically. Transcranial ultrasound was done using LOGIQ P9 ultrasound machine which revealed an open lip schizencephaly involving right cerebral hemisphere with absence of corpus callosum. Trans abdominal sonography revealed non visualisation of left kidney in left renal fossa as well as in its usual ectopic position. Therefore, the new-born underwent magnetic resonance imaging (MRI) of brain and whole abdomen which confirmed ultrasound findings. MRI additionally revealed there was no ocular, anorectal and genito-urinary malformations. Infantogram was done to evaluate the skeleton of this new-born. Fetal echocardiography revealed no obvious cardiac anomalies.

CLINICAL DIAGNOSIS

Except macrocephaly, absent right thumb and deformed left thumb, clinical examination revealed no other abnormalities in this new-born. The mother had no history of diabetes, hereditary disorder, and any potential harmful medicine intake during pregnancy. Infantogram revealed absent phalanges in right thumb and deformed phalanges in left thumb. No abnormalities were noted in remaining skeleton as well as chest of this baby.

Transcranial ultrasound revealed open lip schizencephaly involving right cerebral hemisphere with absence of corpus callosum whereas trans-abdominal sonography revealed non visualisation of left kidney in renal fossa as well as in its usual ectopic position. Ultrasonography (USG) revealed no other abnormalities in this new-born except sludge filled gall bladder. Echocardiography of this new-born revealed no obvious abnormalities. Ocular and auditory examinations revealed no obvious abnormalities. Fetal echocardiography revealed no detectable cardiac anomalies.

Corresponding Author:
Dr. Mojahid Mondal,
Vill - Joyrampur Mondalpara,
PO – Jangipur, PS – Raghunathganj,
Murshidabad - 742213,
West Bengal, India.
E-mail: mojahidmondal69@gmail.com

DOI: 10.18410/jebmh/2021/128

How to Cite This Article:
Mondal M, Pandit N. Acro-renal syndrome with open lip schizencephaly – a rare case report. J Evid Based Med Healthc 2021;8(11):653-656. DOI: 10.18410/jebmh/2021/128

Submission 20-11-2020,
Peer Review 27-11-2020,
Acceptance 21-01-2021,
Published 15-03-2021.

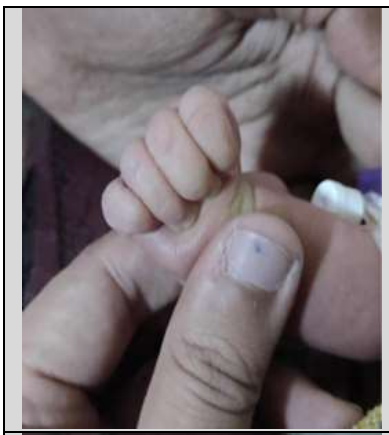

Figure 1.
Absence of Right Thumb

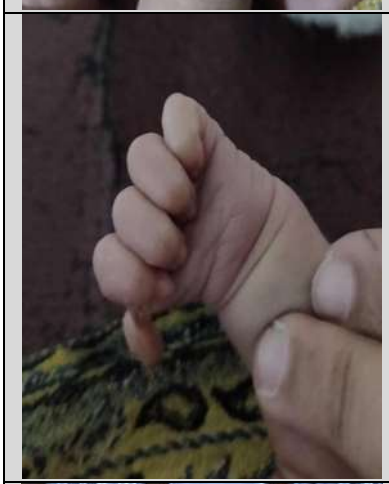

Figure 2.
Malformed Left Thumb

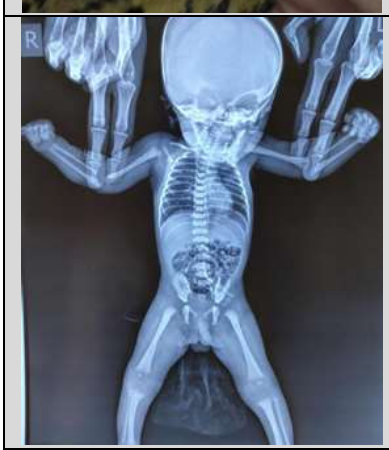

Figure 3.
Infantogram Shows Absence Phalanges in Right Thumb and Hypoplastic Phalanges in Left Thumb. No other Skeletal and Chest Abnormalities Seen

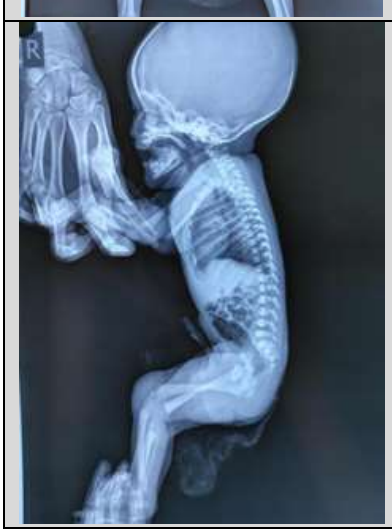

Figure 4.
Lateral Radiograph of Spine Reveals No Obvious Abnormalities

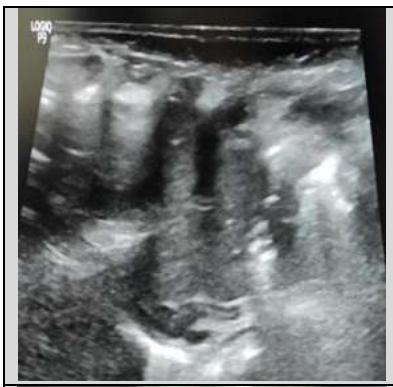

Figure 5.
Coronal USG of Left Hypochondrium Reveals Non-Visualisation of Left Kidney in Left Renal Fossa

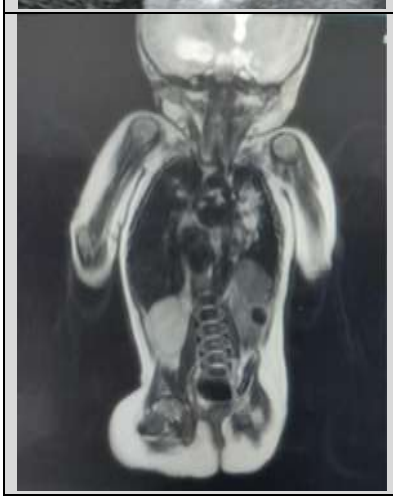

Figure 6.
Coronal MRI Reveals Non-Visualisation of Left Kidney in Left Renal Fossa as Well as in Its Usual Ectopic Position

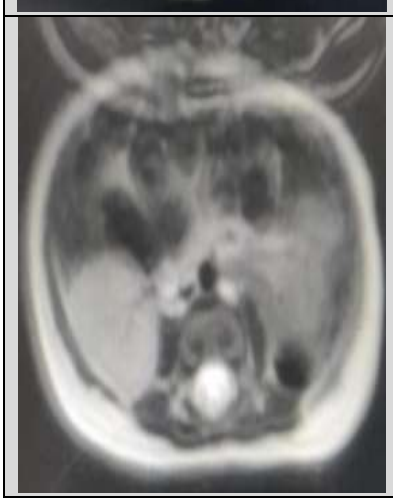

Figure 7.
Axial MR Shows Right Kidney in Its Normal Position with Absence of Left Kidney in Left Renal Fossa

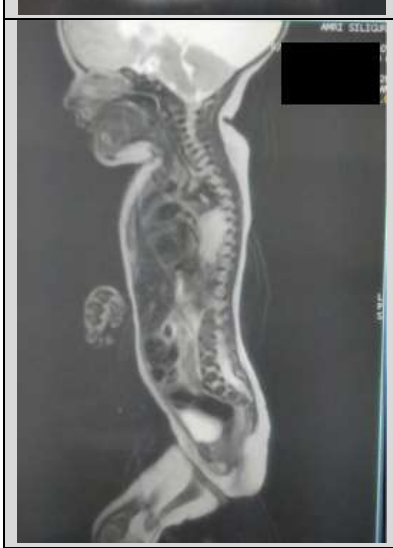

Figure 8.
Sagittal MR of This New-Born Reveals Non-Visualisation of Left Kidney. No Anorectal Abnormalities Seen

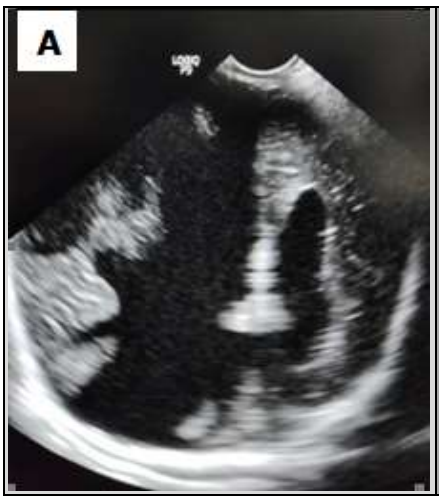

Figure 9A. Transcranial Axial Sonography of Brain Shows Open Lip Schizencephaly

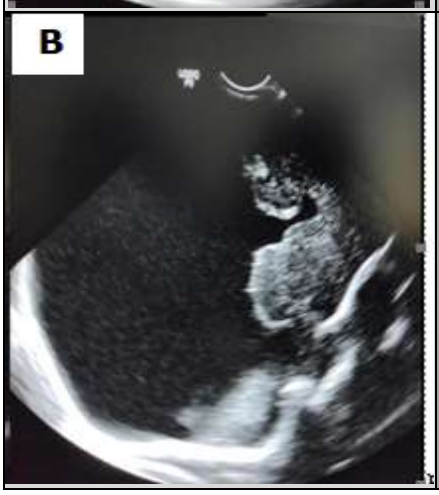

Figure 9B. Transcranial Sagittal Sonography Shows Absence of Corpus Callosum

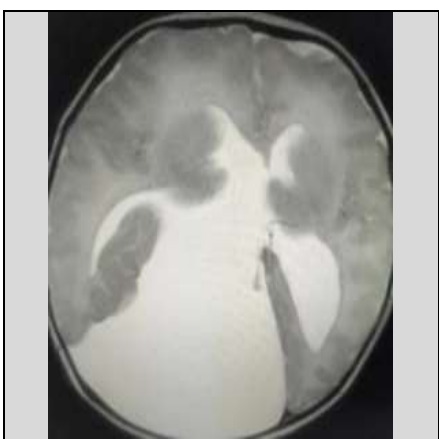

Figure 10. Axial MRI Shows Open Lip Schizencephaly

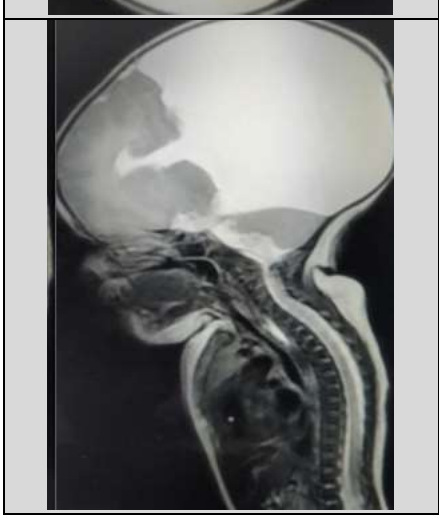

Figure 11. Sagittal MRI Confirms Agenesis of Corpus Callosum

MRI confirmed the USG findings of brain and whole abdomen more decisively and provided additional information of orbit, anorectal and genito-urinary systems. We did not have a facility to do mutational study. So, we considered various radiological findings to achieve a diagnosis. Based on above Clinico-radiological study, we labelled this as a case of acro renal syndrome with open lip schizencephaly.

DIFFERENTIAL DIAGNOSIS

Due to non-availability of definite diagnostic tests or clinical criteria, diagnosis of acro-renal syndrome depends mainly on exclusion of other syndromes based on various prominent clinical manifestations. Various closest differential diagnosis with their brief clinical features are given below:

1. Acro-renal-ocular syndrome: It include upper limb anomalies, mainly thumb hypoplasia, eye abnormalities such as Coloboma and Duane anomaly and renal migration defects.[1]
2. Acro-renal-mandibular syndrome: Major findings are unusual limb deficiencies, renal anomalies and mandibular hypoplasia.[2]
3. Fanconi anaemia: Main features of this rare inherited bone marrow failure syndrome are skin discolorations, hand-arm-other skeletal anomalies, kidney anomaly, small head or eyes, low birth weight, gastrointestinal anomalies, small reproductive organ in males and heart defects.[3]
4. VACTERL association: It includes vertebral abnormalities, anal atresia, cardiac defects, tracheo-oesophageal abnormalities, renal and radial abnormalities, limb abnormalities.[4]
5. Holt Oram syndrome: It is defined as skeletal abnormalities and mild to severe congenital heart defects.[5]
6. Townes-Brocks syndrome: It is a rare autosomal dominant disease characterised by renal, anal, ear and thumb abnormalities.[6]
7. Goldenhar syndrome: It is a developmental disorder characterised by craniofacial anomalies in association with vertebral, cardiac, renal and central nervous system defects.[7]
8. Pallister-Hall syndrome: Includes imperforate anus, renal anomalies (renal hypoplasia or agenesis), limb anomalies (polydactyly, short limbs, syndactyly and nail dysplasia) and genito-urinary abnormalities.[8]

PATHOLOGICAL DISCUSSION

Acro-renal syndrome refers to co-occurrence of congenital renal and limb anomalies. The term acro-renal syndrome was coined by Curran et al. In 1972 though Dieker and Opitz were first to report this phenomenon in three male patients in 1969. The common limb defects include oligodactyly, ectrodactyly, syndactyly of the carpal and tarsal bones, whereas common renal anomalies observed are unilateral renal agenesis, bilateral renal hypoplasia,

ureteric hypoplasia, hydroureteronephrosis and duplication anomalies. The acro-renal syndrome as originally described is rare, reported only in 20 patients in the international literature.[9]

The acro-renal syndrome of Dieker's type is sporadic in nature. The anomalies include unilateral renal agenesis, ectopic kidney, urethral diverticulum, hydroureteronephrosis, ectrodactyly, oligodactyly and hypoplastic carpal / tarsal bones.[10,11]

Acro-renal syndrome of Johnson-Munson is characterised by unilateral or bilateral renal agenesis, aphalangy, hemi vertebrae and genital or intestinal dysgenesis.[12] The acro-renal syndrome of Siegler is characterised by renal ectopia, hydroureteronephrosis, ureteric atresia, short stature, hypoplastic radius / ulna and oligodactyly.[10] The mode of inheritance and gene loci involved in acro-renal syndromes remain unclear, but it is believed they follow an autosomal recessive fashion. A candidate gene for acro-renal disorders is the Formin gene that has been mapped to 15 q 13 - 14. Renal failure reported in patients with acro-renal syndrome is due to oligomeganephronic renal hypoplasia,[13-15] bilateral renal agenesis, secondary focal segmental glomerulosclerosis[16] or other associated urological anomalies.

Open lip schizencephaly is a rare cortical malformation with an estimated incidence of 1.5 / 100000 live births. It is almost always sporadic, although a few familial cases have been described. In which, heterozygous germline mutations of the *homeobox gene EMX2* are often encountered.[17] There is no gender predilection.[17] In open lip schizencephaly, the cleft walls are separated and filled with cerebrospinal fluid (CSF).[17] It is frequently associated (50 – 90 %)[17] with other cerebral anomalies like septo-optic dysplasia, grey matter heterotopia, absent septum pellucidum and dysgenesis of the corpus callosum.[18]

DISCUSSION OF MANAGEMENT

First of all, clinician should identify all anomalies associated with this syndrome to calculate whether it is mild, moderate or severe. Due to very few reported cases, management depends upon various anomalies; approach should be taken accordingly.

We did not know about the genetical inheritance of our case, however, prenatal counseling and early antenatal ultrasonography is necessary to avoid such scenario. In our case, management depends on schizencephaly. Treatment generally consists of physical therapy and drugs to prevent seizures. A surgically implanted tube can be used to shunt the hydrocephalus to another area of body.

FINAL DIAGNOSIS

After a complete clinico-radiological study, we decided this as a case of acro-renal syndrome with open lip schizencephaly and may be designated as a new syndrome.

REFERENCES

[1] Aalfs CM, van Schooneveld MJ, van Keulen EM, et al. Further delineation of the acro-renal-ocular syndrome. Am J Med Genet 1996;62(3):276-281.

[2] Evans JA, Phillips S, Reed M, et al. Severe acro-renal-uterine-mandibular syndrome. Am J Med Genet 2000;93(1):67-73.

[3] Tischkowitz MD, Hodgson SV. Fanconi anaemia. J Med Genet 2003;40(1):1-10.

[4] Solomon BD. VACTERL/VATER association. Orphanet J Rare Dis 2011;6:56.

[5] Holt M, Oram S. Familial heart disease with skeletal malformations. Br Heart J 1960;22(2):236-242.

[6] Kohlhase J. Townes-Brocks Syndrome. In: Genereviews at Genetests: Medical Genetics Information Resource, University of Washington, Seattle. 1997-2007. (Posted 1/24/07). http://www.genetests.org accessed 3/07.

[7] Ashokan CS, Sreenivasan A, Saraswathy GK. Goldenhar syndrome-review with case series. J Clin Diagn Res 2014;8(4):ZD17-ZD19.

[8] Hayek F. Pallister-hall syndrome with orofacial narrowing and tethered cord: a case report. J Med Case Reports 2018;12(1):354.

[9] Natarajan G, Jeyachandran D, Subramaniyan B, et al. Congenital anomalies of kidney and hand: a review. Clin Kidney J 2013;6(2):144-149.

[10] Evans JA, Vitez M, Czeizel A. Patterns of acrorenal malformation associations. Am J Med Genet 1992;44(4):413-419.

[11] Zeier M, Ritz E. No stinkfinger and renal failure - do you see a link? Nephrol Dial Transplant 1999;14:2763-2765.

[12] Curran AS, Curran JP. Associated acral and renal malformations: a new syndrome? Pediatrics 1972;49(5):716-725.

[13] Cascio S, Paran S, Puri P. Associated urological anomalies in children with unilateral renal agenesis. J Urol 1999;162(3 Pt 2):1081-1083.

[14] Kroes HY, Olney RS, Rosano A, et al. Renal defects and limb deficiencies in infants: is it possible to define the acrorenal syndrome? Am J Med Genet 2004;129A(2):149-155.

[15] Akl K. Acrorenal syndrome in a child with renal failure. Am J Med Genet 1994;49(4):447.

[16] Zeier M, Tariverdian G, Waldherr R, et al. Acrorenal syndrome in an adult - presentation with proteinuria, hypertension and glomerular lesions. Am J Kidney Dis 1989;14(3):221-224.

[17] Sarnat HB. Malformations of the nervous system. Elsevier Science Health Science Div 2008.

[18] Donnelly L. Pediatric imaging: the fundamentals. Philadelphia, PA: Saunders/ Elsevier Publication 2009.