Dandy walker variant an association with Rubinstein Taybi syndrome

N Pandya, G Rebello, C Deshpande

Citation

N Pandya, G Rebello, C Deshpande. *Dandy walker variant an association with Rubinstein Taybi syndrome*. The Internet Journal of Pediatrics and Neonatology. 2008 Volume 10 Number 1.

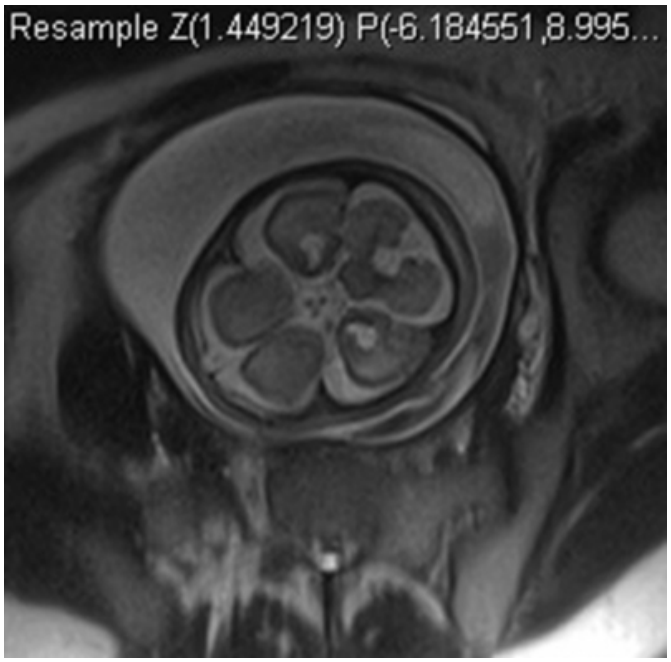
Abstract

Rubinstein Taybi syndrome is a rare malformative syndrome characterised by dysmorphic features and mental retardation. In the neonatal period the diagnosis can be facilitated by the presence of broad thumbs and great toes. Psychomotor and social retardation is present in most of the patients.Most cases are sporadic. The present case has all the main characteristics of Rubin- Taybi syndrome associated with dandy walker variant diagnosed on antenatal MRI as well as post natal Ct scan.

CASE REPORT

A female first born infant was delivered at term by caesarean section. She was found to have a Dandy walker variant on the antenatal scans and later was confirmed by the foetal MRI.

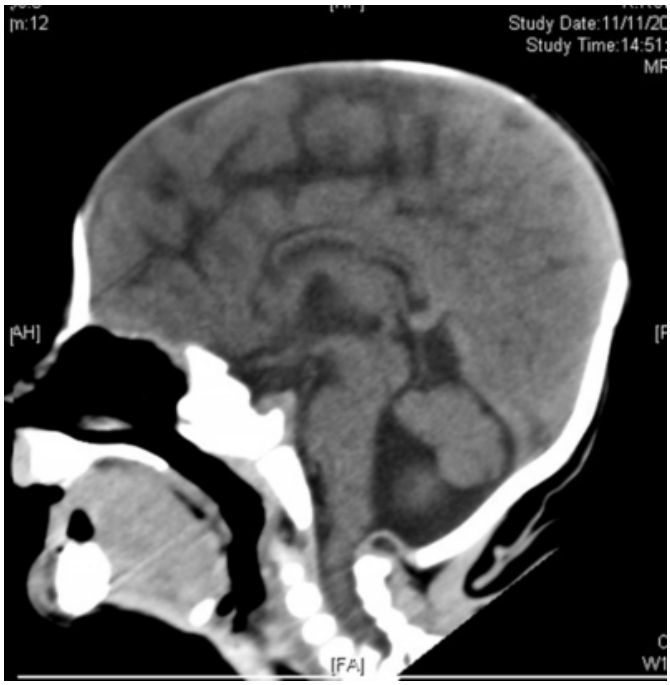
Figure 1



Cystic dilatation with agenesis of Cerebellar vermis.

Post natal Ct confirmed the same.

Figure 2



Cerebellar vermis agenesis with cystic dilatation

Mother declined all genetic screening.

A term neonate born to a primigravida by caesarean section was found to have clinical features consistent with Rubinstein Taybi syndrome. She weighed 2.8 kg had a head circumference of 32 cms .She was found to have hypertrichosis, low hair line, marked hypertelorism, short filtrum ,puffy eye lids and broad toes and thumbs.

Figure 3

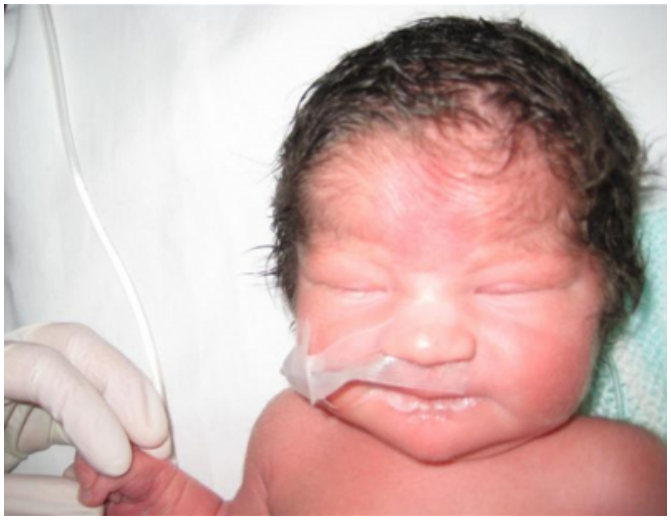
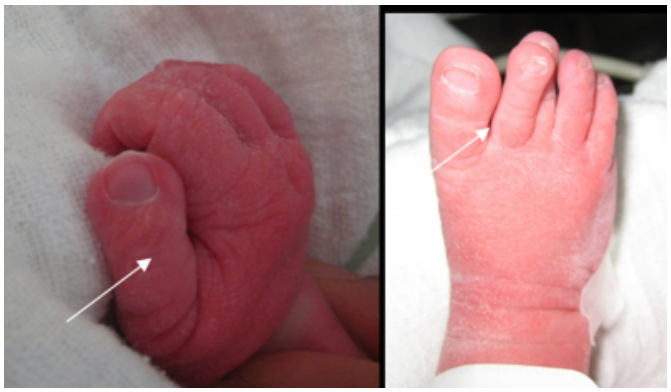


Figure 4



Broad Thumbs and Toes

Her karyotype was normal. The multiplex ligation dependent probe amplication did not reveal any deletion. The CREB p genetic deletion result is still awaited.

DISCUSSION

This syndrome was described first by Rubinstein and Taybi in 1963, characterised by growth and developmental retardation, broad thumbs and large toes and a typical facies. The common abnormalities include hypertrichosis and hirsuitism,heavy eyebrows, prominent beaked nose,hypoplastic maxilla, lowest ears and skeletal anomalies including broad thumbs and large toes,cryptorchidism and cardiac malformations. A neonatal diagnosis is often clinched by the presence of the broad thumbs and toes.[1]

Isolated DW malformation is generally considered nonspecific developmental anomaly. It can occur as a component manifestation of Mendelian or chromosomal syndromes.[2] Broad thumbs and halluces are considered as essential finding of the RTS.The cause is found to be a micro deletion at 16p13.3.[3]

A region of chromosome band 16p13 that includes a gene encoding a binding protein for CREB protein (i.e., CREBBP or CBP) has been associated with the phenotype of RSTS

Feeding difficulties are common in infancy and, together with the genetically based growth retardation characteristic of this syndrome, often result in a clinical picture of failure to thrive . Respiratory infections and complications due to congenital heart disease are major causes of morbidity and mortality in the first years of life. Developmentally, the milestones in these patients are significantly delayed.[4]

CORRESPONDENCE TO

Dr Nikila Pandya Consultant paediatrician Maidstone hospital Hermitage lane Maidstone ME169QQ Fax +441622224495 Email: npandya2@nhs.net

References

1. Agarwal R, Aggarwal R et al. Dandy-Walker malformation in Rubinstein- Taybi syndrome: a rare association. Clinical Dysmorphology 2002,11:223-224
2. Bonioli E, Bellini C, Stefano A. Unusual association: Dandy-walker like malformation in Rubinstein-Taybi syndrome. American Journal of Medical Genetics 1989 ;33:420-421
3. Bartsch O, Schmidt S, Richter M, et al. DNA sequencing of CREBBP demonstrates mutations in 56% of patients with Rubinstein-Taybi syndrome (RSTS) and in another patient with incomplete RSTS. Hum Genet. 2005;117(5):485-493
4. Cantani A, Gagliesi D. Rubinstein Taybi syndrome. Review of 732 cases and analysis of typical traits. . Eur Rev Med Pharmacol Sci. Mar-Apr 1998; 2(2):81-7.

Author Information

N. Pandya, MD,FRCPch
Department Of Paediatrics, Maidstone hospital

Gleena Rebello, MBBS
Department Of Paediatrics, Maidstone hospital

Charu Deshpande, MD ,FRCPch
Department of Genetics, Guys hospital