

# Dandy walker variant an association with Rubinstein Taybi syndrome

N Pandya, G Rebello, C Deshpande

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## 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.

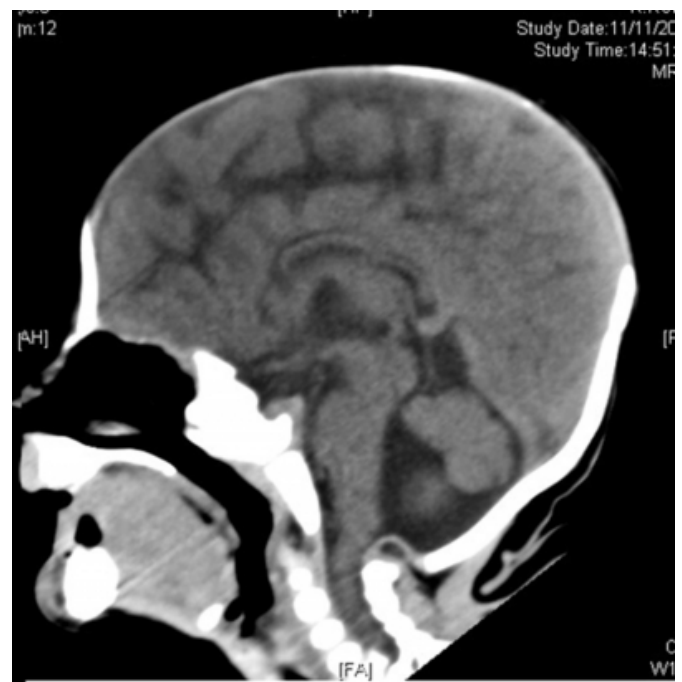
## Figure 1



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 amplification 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

hirsutism, heavy eyebrows, prominent beaked nose, hypoplastic maxilla, low set 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.<sup>1</sup>

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

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.<sup>4</sup>

### **CORRESPONDENCE TO**

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

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**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