

## Case Report

# A case of Ritscher-Schinzel syndrome or 3C syndrome

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**Received:** 26 February 2017

**Accepted:** 28 March 2017

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### ABSTRACT

Ritscher Schinzel syndrome or cranio-cerebello-cardiac syndrome is characterized by cardiac defects, cerebellar hypoplasia and cranial defects. It is usually inherited as autosomal recessive pattern involving chromosome 8q24. the overall prognosis vary widely and it correlates with the cardiac disease present.

**Keywords:** Cardiac septal defect, Enlarged cisterna magna

### INTRODUCTION

Ritscher - Schinzel syndrome (RSS), also known as 3C syndrome, is a clinically heterogeneous disorder characterized by distinctive craniofacial features in addition to cerebellar and cardiac anomalies.

The characteristic central nervous system anomalies are Dandy-Walker malformation, cerebellar vermis hypoplasia or enlargement of the cisterna magna.<sup>1</sup>

The cardiac manifestations include septal and AV canal defects. The cranial dysmorphisms usually seen are a large anterior fontanelle, micrognathia, ocular hypertelorism, brachycephaly, low-set ears, slanted palpebral fissures, cleft palate, depressed nasal bridge and bifid uvula.

### CASE REPORT

A 25 days old female neonate, 2<sup>nd</sup> born to 3<sup>rd</sup> degree consanguineous parents was brought with history of abnormally large head and discolored, prominent eyes.

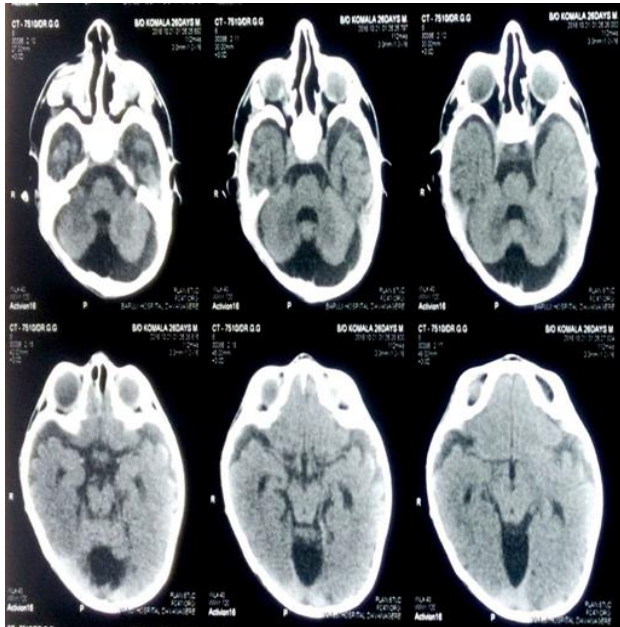
She was born by vaginal delivery with birth weight of 2.3kg. Baby did not have seizures or tone abnormalities.



**Figure 1: Clinical picture of megalocornea, heterochromia iridis, low set ears.**

The infant was on breastfeeds but had poor weight gain. She had macrocephaly with OFC of 39 cm, low set ears, heterochromia iridis with megalocornea (Figure 1), prominent forehead, rib anomalies on left hemithorax, bilateral flat foot. Enlarged cisterna magna with cerebral

atrophy was detected in neuroimaging (Figure 2). Cardiac evaluation revealed a ventricular septal defect with overridden aorta. Ultrasonogram of the abdomen confirmed grade I renal parenchymal disease. A definite history of previous sibling death due to same symptoms was noted. Based on the clinical presentation and imaging studies, diagnosis of Ritscher Schinzel syndrome was made.



**Figure 2: CT Brain with enlarged Cisterna Magna.**

## DISCUSSION

Ritscher - Schinzel syndrome is inherited as autosomal recessive disease and caused by mutation of chromosome 8q24 gene that produces the protein Strumpellin which is involved in endosomal transport and cell death processes.<sup>2,3</sup> The clinical presentation includes central nervous system involvement like Dandy-Walker malformation, enlarged cisterna magna, hydrocephalus. Major cardiac anomalies such as ventricular septal defect, atrial septal defect, tetralogy of Fallot, double-outlet right ventricle, hypoplastic left heart, aortic stenosis, pulmonary stenosis is seen. Commonly encountered craniofacial malformations are low-set ears, hypertelorism, depressed nasal bridge, prominent occiput, cleft palate, micrognathia, ocular coloboma, limb

anomalies, renal malformations, heterochromia iridis, absent ribs, single umbilical artery, congenital glaucoma, hemangioma, hemivertebrae.<sup>4</sup> 3C syndrome is very rare, occurring in less than 1 birth per million. To date < 50 cases have been described. The syndrome appears to be panethnic. Diagnosis is by clinical features and supportive radiological evidence. Management is mainly symptomatic and multidisciplinary approaches including educational programmes, physical, occupational, and speech therapies to improve hypotonia and to reduce motor developmental delay.<sup>5</sup> Cardiac malformations require specific care, often surgery. Prognosis is determined by the cardiovascular and cerebral malformation.<sup>6</sup>

*Funding: No funding sources*

*Conflict of interest: None declared*

*Ethical approval: Not required*

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**Cite this article as:** Deepika G, Raghoji CR, Ashwini RC, Guruprasad G. A case of Ritscher-Schinzel syndrome or 3C syndrome. *Int J Contemp Pediatr* 2017;4:1122-3.