

# CASE REPORT

---

## A RARE ASSOCIATION OF BARDET BIEDL SYNDROME WITH VERTICAL ORBITAL DYSTOPIA AND SITUS INVERSUS WITH DEXTROCARDIA

A. P. R. Naidu<sup>1</sup>, V. Murali Krishna<sup>2</sup>, L. Srijana<sup>3</sup>

### HOW TO CITE THIS ARTICLE:

A. P. R. Naidu, V. Murali Krishna, L. Srijana. "A Rare Association of Bardet Biedl Syndrome with Vertical Orbital Dystopia and Situs Inversus with Dextrocardia". *Journal of Evidence based Medicine and Healthcare*; Volume 2, Issue 7, February 16, 2015; Page: 898-901.

**ABSTRACT:** Bardet Biedl syndrome includes presence of retinitis pigmentosa along with obesity, polydactyl, hypogonadism, renal deformities, and learning disabilities. Orbital dystopia is any type of abnormal displacement of the entire orbital cones and their contents that can occur in three different dimensional planes. Situs describes the anatomic position of cardiac atria and viscera. Situs solitus is normal anatomic position and situs inversus is the mirror image of the situs solitus. We report a case of a 13 year old male whom we diagnosed as a case of BBS with rare association with vertical orbital dystopia and situs inversus with dextrocardia.

**KEYWORDS:** Bardet Biedl syndrome, Situs Inversus, Orbital Dystopia, Dextrocardia.

**INTRODUCTION:** Bardet Biedl syndrome is an autosomal recessive disease which includes systemic features of obesity, polydactyl, hypogonadism, renal deformities and learning disabilities. Ophthalmic manifestations include rod-cone dystrophy, keratoconus, ophthalmoplegia, cataract, ptosis, nystagmus and strabismus.

Facial dysmorphism is present in upto 70% of the diagnosed cases of BBS still no specific set of deformities are consistent, despite some authors describing a subset of children to have facial similarities comprising of deep set eyes, hypertelorism with downward slanting palpebral fissures, a flat nasal bridge with anteverted nares and prominent nasolabial folds, a long philtrum, and a thin upper lip. Presence of a bony deformity like orbital dystopia in association with BBS has only rare if no mention in literature.

**CASE REPORT:** A thirteen year old male presented with ptosis of right lid and progressive night blindness. He was a product of full term normal delivery born out of second degree consanguineous marriage. He has a younger male sibling. He had no family history of RP or any other hereditary disease was diagnosed in the family. He has multiple systemic abnormalities including facial asymmetry due to orbital dystopia, dextrocardia with situs inversus, hypogonadism, fetal lobulation of kidneys, central polydactyl of right hand and learning disabilities. He had moderate ptosis of right upper lid and jaw winking is positive. He had a vertical orbital dystopia of 6mm. Rest of the anterior segment examination is normal for both eyes. The corrected visual acuity of right eye is 6/36 with -10.00 sphere and that of the left eye is 6/36 with -8.00 sphere.

Indirect ophthalmoscopy revealed myopic discs, attenuated vessels, clumps of pigment along the vessels in the mid peripheral retina characteristic of RP. Peripheral constriction of visual fields is observed.

# CASE REPORT

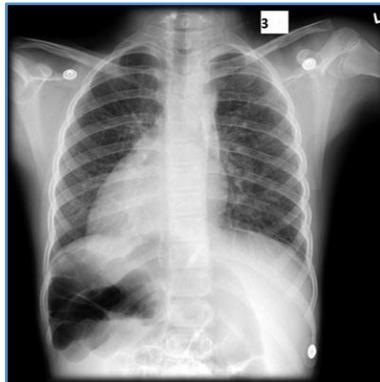
The younger sibling's ocular and systemic examination is within normal limits. His intellectual, and physical development is in accordance with his age.



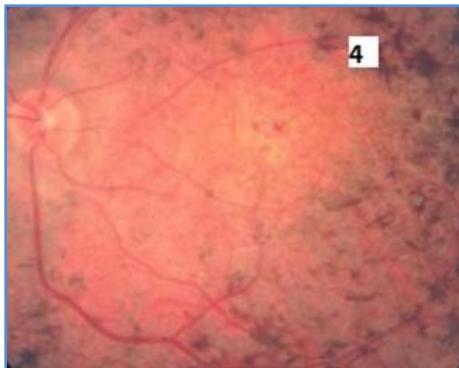
**Fig. 1: Dystopia is clearly seen in this image**



**Fig. 2: Comparative images of the siblings**



**Fig. 3: X-ray film showing situs inversus**



**Fig. 4 & 5: Fundus images showing retinitis**

# CASE REPORT

---

**DISCUSSION:** Bardet-Biedl syndrome is an autosomal recessive and genetically heterogeneous ciliopathy characterized by retinitis pigmentosa, obesity, kidney dysfunction, polydactyly, behavioral dysfunction, and hypogonadism (summary by Beales et al.,).

This syndrome can be quite crippling for the patient. A holistic approach is needed for an early diagnosis and management of this disease. The child we examined, as of now had no metabolic abnormalities, heart, kidney, liver and endocrinal parameters are within normal limits. We need to follow up the child and look out for any newly evolving medical or surgical conditions as the BBS phenotype evolves slowly throughout the first and second decades of life.

Since the child had atypical associations with situs inversus and orbital dystopia, we should be prepared to discover any other atypical features in the child in future.

Another point to stress upon is the follow up of the apparently normal younger sibling keeping in view the consanguineous nature of their birth and atypical presentations seen in the older sibling.

## REFERENCES:

1. Elizabeth Forsythe and Philip L Beales: Bardet–Biedl syndrome (Eur J Hum Genet. Jan 2013; 21(1): 8–13).
2. Lorda-Sanchez, I., Ayuso, C., Ibanez, A Situs inversus and Hirschsprung disease: two uncommon manifestations in Bardet-Biedl syndrome.((Letter) Am. J. Med. Genet. 90: 80-81, 2000)
3. P. L. Beales<sup>a</sup>, N Elcioglu<sup>b</sup>, A S Woolf<sup>c</sup>, D Parker<sup>d</sup>, F A Flinter<sup>a</sup> New criteria for improved diagnosis of Bardet-Biedl syndrome: results of a population survey (J Med Genet 1999; 36: 437-446 doi: 10.1136/jmg.36.6.437).
4. 4.Andrade LJ<sup>1</sup>, Andrade R, França CS, Bittencourt AVPigmentary retinopathy due to Bardet-Biedl syndrome: case report and literature review.(Arq Bras Oftalmol. 2009 Sep-Oct;72(5): 694-6.)
5. OMIM entry # 20990.
6. Moore, S. J., Green, J. S., Fan, Y., Bhogal, A. K., Dicks, E., Fernandez, B. A., Stefanelli, M., Murphy, C., Cramer, B. C., Dean, J. C. S., Beales, P. L., Katsanis, N., Bassett, A. S., Davidson, W. S., Parfrey, P. S. Clinical. And genetic epidemiology of Bardet-Biedl syndrome in Newfoundland: a 22-year prospective, population-based, cohort study (Am. J. Med. Genet. 132A: 352-360, 2005.0).

# CASE REPORT

---

**AUTHORS:**

1. A. P. R. Naidu
2. V. Murali Krishna
3. L. Srijana

**PARTICULARS OF CONTRIBUTORS:**

1. Associate Professor, Department of Ophthalmology, Rangaraya Medical College.
2. Professor & HOD, Department of Ophthalmology, Rangaraya Medical College.
3. Junior Resident, Department of Ophthalmology, Rangaraya Medical College.

**NAME ADDRESS EMAIL ID OF THE CORRESPONDING AUTHOR:**

Dr. A. P. R. Naidu,  
Assistant Professor,  
Department of Ophthalmology,  
Rangaraya Medical College/GGH,  
Kakinada-533001.  
E-mail: aprinaidu2@gmail.com

Date of Submission: 02/02/2015.  
Date of Peer Review: 03/02/2015.  
Date of Acceptance: 04/02/2015.  
Date of Publishing: 13/02/2015.