Case Report

# Laurence moon bardet beidl syndrome

Ajit A. Holkunde[1*], Anant K. Shingare[2], Supriya B. Sakhare[3], D.S. Joshi[4]

[1]Post-Graduate student, [**]Assistant Professor, Department of Anatomy, SRTR Government Medical College, Ambajogai-431517, Maharashtra, INDIA.
[4]Professor and Head, Department of Anatomy, Dr. Shankarrao Chavan Government Medical College, Nanded, Maharashtra, INDIA.
**Email:** drholkunde@gmail.com

## Abstract

**Introduction:** A 15 year old female patient was brought by the parents to the Out-Patient department of medicine of our institute, SRTR Medical College on 11th of Nov, 2011. The presenting complaints were- Dyspnoea since 15 days, Fatigue since 15 days. On general examination, it was found that patient was having severe pallor, truncal obesity as well as mental retardation. At the same time, one more characteristic finding was quadrilateral polydactyly. Her systemic examination of CVS and RS was normal. On fundoscopy, positive finding was retinitis pigmentosa. All these clinical signs and symptoms direct us towards the diseases of genetic origin, to be more precise, towards Laurence Moon Bardet Beidl syndrome or Prader-Willi syndrome. Karyotype of this patient may give additional support to the diagnosis.
**Keywords:** Truncal obesity; quadrilateral polydactyly; retinitis pigmentosa; Prader-Willi syndrome; karyotype;

[*]**Address for Correspondence:**
Dr. Ajit A. Holkunde, Department of Anatomy, SRTR Government Medical College, Ambajogai-431517, Maharashtra, INDIA.
**Email:** drholkunde@gmail.com
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## INTRODUCTION

The world is full of things which always amaze the human beings. But the fact that human being itself is full of such surprising features. That makes the study of human development important. Any deviation in this normal path of development gives rise to the syndromes with varying presenting features. Laurence Moon Bardet Beidl Syndrome, first described by Bardet[1] in 1922, is one of these syndromes. It is an autosomal recessive syndrome characterised by functional and structural abnormalities of organs and tissues with diverse embryonic derivation. World over, the disease is rare, affecting only 1in 160,000 people[2]. However, it shows a higher rate of prevalence in the Arab population. Diagnosis depends entirely on the clinical features. The five cardinal features include polydactyly or syndictily, pigmentary retinopathy, obesity, mental retardation and hypogonadism. Patient usually presents in first 10 years of life with complaint of poor vision at night. Most patients show the ophthalmological complaints and are legally blind at the age of 30 yrs. Other systemic features include brachycephaly, short stature, congenital heart block, deafness and kidney disorders.[3,4,5]

## MATERIAL AND METHODS

A 15 yr old female patient, residing at a village in taluka parli of district beed, came with the chief complaints of dyspnoea since 15 days, fatigue since 15 days. She was also complaining of low vision in darkness. On general examination, patient was obese. One characteristic finding was polydactyly of all four limbs. Severe pallor was also noted. Other findings on examination were short stature, mental retardation. Systemic examination showed normal RS, CVS. Menstrual history found primary amenorrhea, breasts were at Tanner stage II. Family history was normal. On fundoscopy, retinal pigmentation with disc pallor were noted. USG abdomen and pelvis was normal. CBC showed Hb level of 7gm%

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Figure 1

## DISCUSSION

Laurence moon bardet beidl syndrome is classified in two classes-one is Laurence Moon syndrome, characterised by spastic paraplegia and absence of polydactyly, obesity, which are the features of second class i.e. Bardet Beidl Syndrome. But linkage analysis have shown both the diseases to be linked with Chr11q region[2]. The above patient shows the feature of Bardet Beidl syndrome. Prader-willi syndrome is another similar syndrome characterised by obesity, mental retardation and neonatal hypotonia. This can be differentiated from LMBB syndrome because absence of polydactyly, hypotonia. Reason for obesity in prader-willi syndrome is overeating. Prader- Willi syndrome is caused by the lack of functioning paternally- inherited genes in critical regions of the Chr15. Failure to elicite family history is not surprising as in case of Mendelian recessive type[6]. In the LMBB syndrome, though the diagnosis entirely done on the basis of clinical grounds, karyotype may give additional support. But because of nonco-operative nature of patient, karyotype was not possible.

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