http://www.banglajol.info/index.php/JACEDB

Case report

DOI: https://doi.org/10.3329/jacedb.v5i1.86707

Journal of Association of Clinical Endocrinologist and Diabetologist of Bangladesh
January 2026, Vol. 5, No. 1, pp. 77-80

ISSN (Online): 2959-8176
ISSN (Print): 2958-0307

Berardinelli–Seip Congenital Lipodystrophy in a child: Classical phenotype with multisystem involvement from a resource-limited setting

*Banik JD[1], Hossain MA[2], Mondal E[3], Banik SR[4]

[1]Joy Datta Banik, Post-graduate student, Department of Endocrinology, Mymensingh Medical College, Mymensingh, Bangladesh;
[2]Md. Anowar Hossain, Professor, Department of Endocrinology, Mymensingh Medical College, Mymensingh, Bangladesh;
[3]Ershad Mondal, Assistant Professor, Department of Endocrinology, Mymensingh Medical College, Mymensingh, Bangladesh;
[4]Sanjoy Ranjon Banik, Medical Officer, Department of Endocrinology, Mymensingh Medical College, Mymensingh, Bangladesh

Abstract

Berardinelli–Seip Congenital Lipodystrophy (BSCL) is a rare autosomal recessive disorder characterized by near-total absence of adipose tissue and severe metabolic derangements beginning in early life. We report a 10-year-old boy presenting with generalized lack of subcutaneous fat, acromegaloid features, hepatomegaly, insulin resistance, dyslipidemia, and hypertrophic cardiomyopathy. Characteristic clinical features supported by biochemical and radiological investigations established the diagnosis of BSCL. Genetic confirmation could not be performed due to limited resources. This case emphasizes the importance of careful phenotypic recognition for early diagnosis and timely management of BSCL, particularly in low-resource settings, to prevent long-term metabolic and cardiovascular complications. Early phenotypic recognition is critical for diagnosis of BSCL in the absence of genetic testing; severe insulin resistance and hepatic involvement are key diagnostic clues; comprehensive multisystem evaluation is essential in affected children. *[J Assoc Clin Endocrinol Diabetol Bangladesh,* January 2026; 5(1): *77-80]*

Keywords: Berardinelli-Seip syndrome; Congenital generalized lipodystrophy; Insulin resistance; Hepatomegaly; Pediatric metabolic disorder; Resource-limited setting

***Correspondence:** Dr. Joy Datta Banik, Post-graduate trainee, Department of Endocrinology, Mymensingh Medical College, Mymensingh, Bangladesh Email: jdbanik82@gmail.com, Phone: 01786698744

Introduction

Berardinelli-Seip Congenital Lipodystrophy (BSCL), also known as congenital generalized lipodystrophy, is a rare autosomal recessive disorder characterized by near-complete absence of adipose tissue from birth or early infancy. The condition leads to severe insulin resistance, hypertriglyceridemia, hepatic steatosis, early-onset diabetes mellitus, and cardiomyopathy.[1-4] BSCL results from mutations in genes involved in adipocyte differentiation and lipid storage, leading to failure of normal adipose tissue development and ectopic fat deposition.[5] Owing to its rarity and variable presentation, diagnosis is often delayed, especially in resource-limited settings where genetic testing is unavailable. Early recognition is crucial to reduce metabolic and cardiovascular morbidity.

Case report

A 10-year-old boy presented with progressive abdominal distension, poor weight gain, and abnormal facial appearance since early childhood (Figure-1). He was born at term following an uncomplicated pregnancy and delivery. The parents were non-consanguineous. There was no family history of similar illness, diabetes, cardiomyopathy, or early deaths, and no history of neonatal hypoglycemia.

On physical examination, the child had a striking generalized lack of subcutaneous fat with prominent musculature and superficial veins. Facial features were acromegaloid with prognathism, enlarged hands and feet, and coarse facial appearance. The abdomen was protuberant with firm hepatomegaly. Growth parameters revealed weight and height below the age-appropriate percentiles, with body mass index significantly reduced

for age, despite preserved muscular bulk. Blood pressure was within normal range for age.

Laboratory investigations demonstrated fasting hyperglycemia, markedly elevated fasting insulin levels, hypertriglyceridemia, and deranged liver enzymes (Table-I). Homeostatic model assessment showed severe insulin resistance and impaired β-cell function. HOMA-IR and HOMA-B were calculated using standard formulas and demonstrated marked insulin resistance with β-cell dysfunction. Serum leptin estimation and

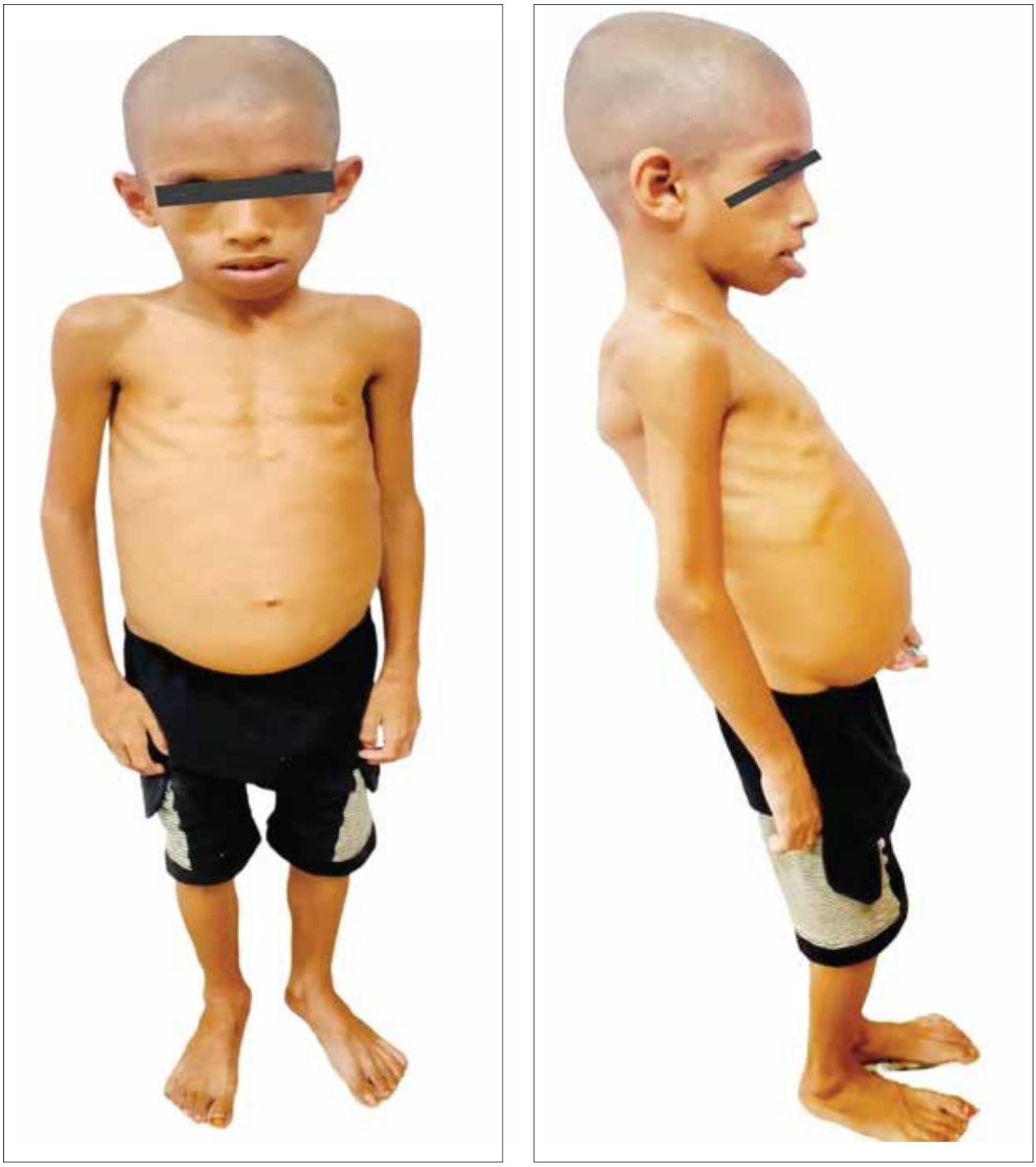

Figure-1: A 10-year-old male having generalized lipodystrophy, acromegaloid facies, and upper abdominal distension

Table-I: Laboratory investigations of the patient

Name of investigations	Result	Reference value
FPG (mmol/L)	19.2	3.5-5.5
PPG (mmol/L)	20.8	<7.8
HbA1c (%)	11.8	4-5.6
C- Peptide (ng/mL)	1.56	0.78-5.19
Serum Sodium (mmol/L)	135.7	135-148
Serum Potassium	3.88	3.5-5.5
Serum Chloride	100	98-108
Serum Bicarbonate	24.7	22-28
Urine R/E	Sugar (+++), Albumin-Nil	-
CRP (mg/L)	<6.0	Up to 06
Serum creatinine (mg/dl)	0.6	0.5-1.2
Serum Uric Acid (mg/dL)	3.1	2-7
S. Bilirubin (mg/dL)	0.7	0.1-1.2
SGPT (U/L)	51	<41
HbsAg	Negative	-
Anti HCV	Negative	-
Serum TSH (µIU/ml)	1.46	0.37-6.0
Serum FT4 (fmol/ml)	13.62	8.56-25.6
Serum calcium (mg/dl)	10.0	8.5-10.5
Serum Albumin (gm/dl)	4.4	3.6-5.2
Serum Cholesterol (mg/dl)	171	150-200
LDL-c (mg/dl)	51	<150
HDL-c (mg/dl)	42	35-65
TG (mg/dl)	388	<150
Echocardiogram	Hypertrophic Cardiomyopathy; Mild TR with (PASP = 30 mmHg); Good bi-ventricular function (LVEF: 72%)	
USG of W/A	Hepatomegaly with fatty change	
Liver Biopsy	Compatible with lipid storage disease, progressive to early cirrhosis	
Upper GI Endoscopy	Normal	

FPG: Fasting Plasma Glucose; PPG: Post-Prandial Glucose; HbA1c: Glycated Hemoglobin (Hemoglobin A1c); R/E: Routine Examination; CRP: C-Reactive Protein; SGPT: Serum Glutamic Pyruvic Transaminase; HBsAg: Hepatitis B Surface Antigen; Anti HCV: Antibody against Hepatitis C Virus; TSH: Thyroid Stimulating Hormone; FT4: Free Thyroxine; LDL-c: Low-Density Lipoprotein Cholesterol; HDL-c: High-Density Lipoprotein Cholesterol; TG: Triglycerides; TR: Tricuspid Regurgitation; PASP: Pulmonary Artery Systolic Pressure; LVEF: Left Ventricular Ejection Fraction; USG of W/A: Ultrasonography of Whole Abdomen; GI: Gastrointestinal

genetic testing were not available.

Ultrasonography of the abdomen revealed hepatomegaly with fatty infiltration. Echocardiography showed features of hypertrophic cardiomyopathy with preserved systolic function. Liver biopsy was performed to assess the extent of hepatic involvement and to exclude storage disorders, demonstrating macrovesicular steatosis. Bone marrow examination was undertaken to exclude infiltrative or hematological causes of hepatomegaly and was unremarkable.

Based on characteristic clinical phenotype and supportive metabolic and imaging findings, a diagnosis of Berardinelli–Seip Congenital Lipodystrophy was made.

Discussion

BSCL is a rare disorder with an estimated prevalence of less than 1 per million population.[1] The diagnosis in this patient was established clinically due to the presence of generalized absence of adipose tissue from early childhood, acromegaloid features, severe insulin

resistance, dyslipidemia, hepatic steatosis, and cardiomyopathy.

The differential diagnoses considered included acromegaly, Cushing syndrome, progeroid syndromes, and partial lipodystrophy. Acromegaly was excluded due to the patient's age and absence of pituitary pathology. Cushing syndrome was unlikely in the absence of cortisol excess and typical clinical features. Progeroid syndromes usually present with growth failure and premature aging rather than metabolic derangements. Partial lipodystrophy was excluded due to the generalized and early-onset absence of adipose tissue, which is characteristics of BSCL.[1,3]

BSCL is caused by mutations in genes such as AGPAT2, BSCL2, CAV1, and PTRF, which are essential for adipocyte differentiation, lipid droplet formation, and triglyceride storage.[4-7] Loss of functional adipose tissue results in ectopic lipid deposition in liver, muscle, and myocardium, leading to insulin resistance, hepatic steatosis, and cardiomyopathy.[4,8,9]

Liver biopsy was justified to assess the severity of hepatic involvement and to exclude other causes of hepatomegaly, while bone marrow examination was performed to rule out infiltrative disorders in view of marked organomegaly.[5]

Limitations of this case include the unavailability of genetic confirmation, serum leptin estimation, and advanced therapeutic options such as recombinant leptin therapy, reflecting challenges commonly faced in low-resource settings.[10]

Conclusions

This case highlights the importance of recognizing the characteristic phenotype of Berardinelli–Seip Congenital Lipodystrophy for early diagnosis, even in the absence of genetic testing. Timely identification and comprehensive evaluation are essential to manage metabolic complications and reduce long-term morbidity, particularly in resource-limited settings.

Acknowledgements

We are grateful to the patient and his attendants for consenting to the clinical photography and its publication.

Disclosure

The authors have no conflicts of interest to disclose.

Financial Disclosure

The authors received no specific funding for this work.

Data Availability

Any inquiries regarding supporting data availability of this study should be directed to the corresponding author and are available from the corresponding author upon reasonable request.

Ethical Approval and Consent to Participate

Written informed consent was obtained from the patient's attendant. All methods were performed in accordance with the relevant guidelines and regulations.

How to cite this article: Banik JD, Hossain MA, Mondal E, Banik SR. Berardinelli–Seip Congenital Lipodystrophy in a child: Classical phenotype with multisystem involvement from a resource-limited setting. J Assoc Clin Endocrinol Diabetol Bangladesh, 2026; 5(1):77-80

Publication History

Received on: 27 December 2025

Accepted on: 30 January 2026

Published on: 31 January 2026

References

1. Garg A. Acquired and inherited lipodystrophies. N Engl J Med 2004;350:1220-34. DOI:10.1056/NEJMra025261.
2. Berardinelli W. An undiagnosed endocrinometabolic syndrome. J Clin Endocrinol Metab 1954;14:193-204. DOI: 10.1210/jcem-14-2-193.
3. Seip M, Trygstad O. Generalized lipodystrophy, congenital and acquired. Acta Paediatr Suppl 1996;413:2-28. DOI: 10.1111/j.1651-2227.1996.tb14262.x.
4. Agarwal AK, Garg A. Genetic basis of lipodystrophies and management of metabolic complications. Annu Rev Med 2006;57:297-311. DOI: 10.1146/annurev.med.57.121304.131244.
5. Van Maldergem L, Magré J, Khallouf TE, Gedde-Dahl T Jr, Delépine M, Trygstad O, et al. Genotype-phenotype relationships in Berardinelli-Seip congenital lipodystrophy. J Med Genet 2002;39(10):722-33. DOI: 10.1136/jmg.39.10.722.
6. Magré J, Delépine M, Khallouf E, Gedde-Dahl T Jr, Van Maldergem L, Sobel E, et al. Identification of the gene altered in Berardinelli-Seip congenital lipodystrophy on chromosome 11q13. Nat Genet 2001;28:365–70. DOI:10.1038/ng585.
7. Agarwal AK, Arioglu E, De Almeida S, Akkoc N, Taylor SI, Bowcock AM, et al. AGPAT2 mutations in congenital generalized lipodystrophy. Nat Genet 2002;31:21-3. DOI:10.1038/ng880.
8. Simha V, Garg A. Clinical features and metabolic derangements in acquired generalized lipodystrophy. J Clin Endocrinol Metab 2003;88:543–52. DOI:10.1210/jc.2002-021030.
9. Bjornstad PG, Foerster A, Ihlen H. Cardiac findings in congenital generalized lipodystrophy. Acta Paediatr 1996;85:39-43. DOI:10.1111/j.1651-2227.1996.tb13864.x.
10. Chong AY, Lupsa BC, Cochran EK, Gorden P. Efficacy of leptin replacement therapy in the different forms of human lipodystrophy. Diabetes 2010;59:255–63. DOI:10.2337/db09-0872.