International Journal of Community Medicine and Public Health
Gupta J et al. Int J Community Med Public Health. 2026 May;13(5):2507-2510
http://www.ijcmph.com
pISSN 2394-6032 | eISSN 2394-6040

DOI: https://dx.doi.org/10.18203/2394-6040.ijcmph20261444

Case Report

Hutchinson–Gilford progeria syndrome in a 2-year-11-month-old female child: a clinico-genetic case report from Assam, India

Juhi Gupta*, Sonanki Deb, Suranjita Mazumdar

Department of Psychiatry, Lokopriya Gopinath Bordoloi Regional Institute of Mental Health (LGBRIMH), Tezpur, Assam, India

Received: 25 January 2026
Revised: 24 March 2026
Accepted: 27 March 2026

***Correspondence:**
Dr. Juhi Gupta,
E-mail: juhi31071990@gmail.com

ABSTRACT

Hutchinson–Gilford Progeria Syndrome (HGPS) is a rare genetic disorder characterized by premature aging beginning in early infancy. It is caused by de novo pathogenic variants in the LMNA gene. We report a case of a 2-year-11-month-old female child from Nagaon district of Assam who presented with failure to thrive and progressive craniofacial and somatic changes. Clinical features were classical for Progeria, with preserved cognitive and social development. Whole Exome Sequencing confirmed a pathogenic heterozygous variant in exon 11 of the LMNA gene, consistent with Hutchinson–Gilford Progeria Syndrome. This report highlights early clinical recognition, genetic confirmation, and the psychosocial impact on caregivers, emphasizing the role of counseling and multidisciplinary care.

Keywords: Hutchinson–Gilford progeria syndrome, LMNA gene, Premature aging, Child psychiatry, Caregiver burden

INTRODUCTION

Hutchinson–Gilford Progeria Syndrome (HGPS) is an extremely rare sporadic disorder with an estimated incidence of 1 in 4–8 million live births. It results from de novo mutations in the LMNA gene encoding lamin A, leading to the production of progerin, an abnormal protein responsible for nuclear instability and accelerated cellular aging. Affected children usually appear normal at birth, with phenotypic features emerging within the first few months to years of life.

The disorder is characterized by severe postnatal growth failure, distinctive facial appearance, alopecia, skin atrophy, loss of subcutaneous fat, skeletal abnormalities, and early cardiovascular complications. Intelligence is typically normal. While most published reports focus on the medical aspects of the condition, fewer highlight the psychosocial impact on families and the role of mental health services. We present a genetically confirmed case of HGPS from Northeast India, highlighting clinical features, genetic findings, and caregiver-related concerns.

Review of literature

Research on childhood terminal illnesses such as progeria highlights the complex interplay of medical, psychological, and social factors that shape the experiences of afflicted children and their families. Most studies on the psychosocial management of progeria draw on literature from other childhood life-limiting illnesses, particularly pediatric cancer. Parents often struggle with anxiety, denial, guilt, and anger while confronting the diagnosis and its implications. Studies note a significant gap in research regarding personality attributes and coping strategies among children with progeria, as well as parental grief responses.[1] In qualitative interviews, initial reactions to diagnosis are often described as

psychologically overwhelming, followed by attempts to deny or alter the perceived fatal trajectory through repeated consultations and self-blame. Living with progeria poses emotional challenges not only for the child but also for the entire family.[2] Rehabilitative interventions play a vital role in enhancing quality of life.

Occupational therapy focuses on life skills, adaptive equipment, and fine motor development, often in collaboration with physical therapy for comprehensive care.[3] Children with progeria benefit from simple environmental adaptations to enhance independence and participation, including home modifications (bathroom steps, adaptive tools, lowered surfaces), mobility-friendly spaces, and safe recreational options. Small tablets, mini keyboards, school accommodations, and easy-to-wear clothing further support daily functioning and social inclusion.[3] Psychosocial interventions also address spiritual concerns, family conflict, and caregiver coping skills.[1]

In summary, while medical management of life-limiting childhood conditions has advanced, the psychological burden on children and families remains substantial, necessitating integrated care models that combine clinical, psychological, and developmental perspectives tailored to each child's context.

CASE REPORT

A 2-year-11-month-old female child was brought to the Child and Adolescent Psychiatry outpatient department at Lokopriya Gopinath Bordoloi Regional Institute of Mental Health (LGBRIMH), Tezpur, by her parents with complaints of unusual facial appearance and failure to thrive.

The child belonged to Nagaon district of Assam. She was the first child born to non-consanguineous parents. The mother was a homemaker and the father was a farmer. The family belonged to a lower-middle socioeconomic background. There was no family history of premature aging, genetic disorders, neurological illness, or psychiatric disorders.

Perinatal and developmental history

The child was born by normal vaginal delivery following an uneventful antenatal and perinatal period. There was no history of birth asphyxia, neonatal seizures, or prolonged neonatal intensive care unit stay.

According to the parents, physical changes were first noticed at around 2.5 months of age, in the form of poor weight gain and subtle changes in facial appearance. Over subsequent months, progressive hair thinning, skin changes, and disproportionate growth compared to peers became evident. Despite concerns regarding physical growth, developmental milestones—particularly language, social interaction, and adaptive behaviors—were achieved appropriately for age.

Clinical examination

On physical examination, the child appeared markedly small for her age with significant growth retardation. Anthropometric measurements were as height 62 cm and torso length 28 cm.

Distinctive physical features included

The patient presented with prominent bulging eyes and a disproportionately large head, which progressively increased with age. Additional features included sparse scalp hair with marked thinning, absence of eyebrows, and a beaked nose. Facial and skeletal abnormalities were noted, including micrognathia, while the body showed a protuberant abdomen (pot belly) and thin limbs with reduced subcutaneous fat.

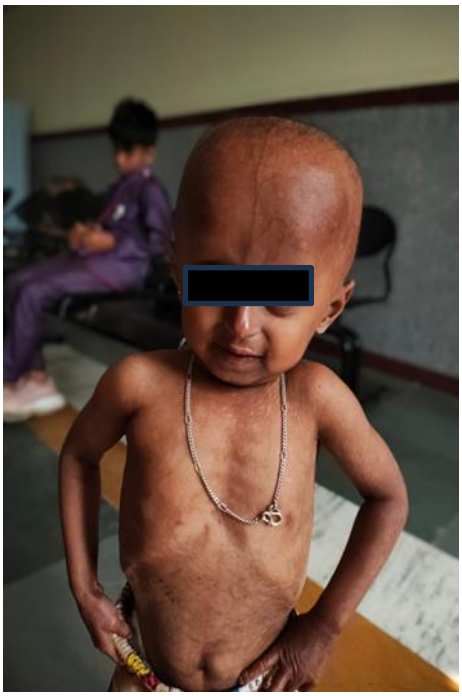

Figure 1: Typical facies of progeria: senile look with prominent eyes, sparse hair, beaked nose with mottled pigmentation over the trunk.

The overall clinical picture was strongly suggestive of HGPS.

Mental status and developmental assessment

The child was alert, cooperative, and socially responsive during the assessment. Eye contact, social reciprocity, and communication were appropriate for age. Assessment using the Vineland Social Maturity Scale (VSMS) revealed a Social Quotient (SQ) of 96, indicating average social and adaptive functioning for chronological age.

Cognitive functioning appeared intact, consistent with existing literature on HGPS.

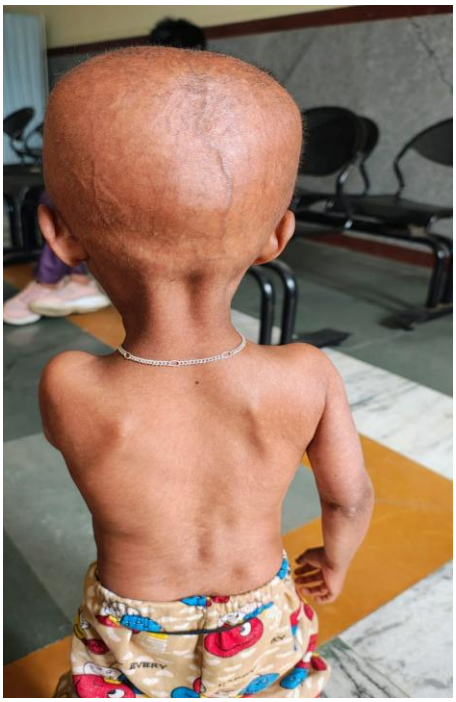

Figure 2: Skin tightening and prominence of veins.

Investigations

Electroencephalogram (EEG) findings were normal, and systemic evaluation revealed no cardiac, neurological, or other comorbidities at the time of assessment. Psychiatric evaluation similarly indicated the absence of any comorbid psychiatric disorder.

Genetic evaluation

Based on clinical suspicion, Whole Exome Sequencing (WES) was advised and performed.

The analysis revealed

Genetic analysis identified a heterozygous variant in exon 11 of the LMNA gene (chromosome 1:g.156138613C>T), corresponding to c.1824C>T (p Gly608=). Although this is a synonymous change, it is known to activate a cryptic splice site, leading to the production of abnormal Lamin A (progerin). This variant has been previously reported in patients with HGPS and is classified as pathogenic in ClinVar. Population frequency data indicate that it is rare or absent in large population databases, and functional studies along with existing literature support its pathogenic role. Based on clinical correlation and genetic findings, the diagnosis of HGPS was confirmed. Parental testing was recommended, considering the mutation is typically de novo.

Psychosocial assessment

Both parents exhibited significant emotional distress and high caregiver burden. There was limited prior awareness about the disorder, its genetic basis, prognosis, and long-term implications. Expressed emotions included anxiety, guilt, and fear regarding the child's future health and survival. Additional concerns related to social stigma, financial constraints, and uncertainty about long-term care needs were reported.

Intervention and counselling

Structured counseling was conducted using the BREAKS protocol. Parents were provided with clear and simple explanations regarding:

The discussion with the family addressed the genetic and sporadic nature of the disorder, outlining the expected disease course and prognosis. Emphasis was placed on the preservation of intelligence and emotional development, as well as the importance of regular follow-up and multidisciplinary care, particularly involving paediatric and cardiology surveillance. Parental concerns were addressed, misconceptions clarified, and reassurance provided to reduce guilt and self-blame. The family was advised to continue follow-up and supportive care.

DISCUSSION

This case represents a classical presentation of HGPS with early onset, characteristic physical features, preserved cognitive development, and significant caregiver distress. The LMNA exon 11 mutation identified is the most commonly reported pathogenic variant globally and is consistent with previously published Indian and international case reports.[1]

An important aspect highlighted is the psychosocial impact on caregivers and the role of mental health professionals in providing counseling, emotional support, and guidance. Early diagnosis allows families to understand the condition, plan care, and access appropriate monitoring and support services.

CONCLUSION

HGPS should be considered in infants and young children presenting with failure to thrive and characteristic craniofacial and dermatological features.

Genetic confirmation aids in definitive diagnosis and counseling. A multidisciplinary approach, including psychiatric support for caregivers, is essential for comprehensive management of this rare condition.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

REFERENCES

1. Livneh H, Antonak RF, Maron S. Progeria: medical aspects, psychosocial perspectives, and intervention guidelines. Death stud. 1995;19(5):433-52.
2. LongLifeNutri. Living with Progeria: Daily Challenges, Support, and New Research Insights. 2023. Available at: https://www.longlifenutri.com/blogs/news/living-with-progeria-daily-challenges-support-and-new-research-insights. Accessed on 25 December 2025.
3. Progeria Research Foundation. Handbook 2018. 2018. Available at: https://www.progeriaresearch.org/wpcontent/uploads/2019/01/Handbook-2018-PDF-One-File-v3-1.pdf. Accessed on 25 December 2025.

Cite this article as: Gupta J, Deb S, Mazumdar S. Hutchinson–Gilford progeria syndrome in a 2-year-11-month-old female child: a clinico-genetic case report from Assam, India. Int J Community Med Public Health 2026;13:2507-10.