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Beyond the Aged Face: A Neonatal Case of Ectodermal Dysplasia

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Abstract

Background:
Ectodermal dysplasia is a rare genetic condition that affects the development of the skin, hair, nails, teeth, and sweat glands. It can be difficult to diagnose in newborns, especially when symptoms resemble other conditions like progeria.

Case Presentation: We present the case of a 1-month-old baby boy who was brought in for poor weight gain and feeding difficulties. On examination, he had sparse hair, a prominent forehead, deep-set eyes, dry and wrinkled skin, and thin nails. His appearance was initially thought to resemble progeria, a premature aging syndrome. However, his routine tests were normal and imaging didn't support this diagnosis. Genetic testing (whole exome sequencing) later confirmed he had hypohidrotic ectodermal dysplasia due to a mutation in the *EDA* gene.

Management and Outcome: He was admitted to the ICU, started on orogastric feeds, and gradually transitioned to oral feeding. Skin care and supportive measures were continued. The baby was discharged in stable condition with plans for regular follow-up and genetic counselling for the family. **Conclusion:** This case highlights the need to consider ectodermal dysplasia in infants with unusual features and poor growth. Early genetic testing helps ensure correct diagnosis and timely care.

Keywords: Anhidrosis, Ectodermal dysplasia, Hypodontia, Hypotrichosis, Neonate

Introduction

Ectodermal dysplasias (EDs) are a heterogeneous group of inherited disorders characterized by abnormalities of ectodermally derived structures, including the skin, hair, teeth, nails, and sweat glands. The estimated incidence of ED is around 1 in 100,000 live births, with **hypohidrotic ectodermal dysplasia** being the most common form [1,2]. Clinical presentation varies with age and severity, and diagnosis in neonates can be particularly challenging due to overlapping phenotypes with other syndromic disorders such as progeria or other genodermatoses [3,4].

Failure to thrive (FTT) in neonates can be a manifestation of various underlying systemic or genetic disorders. Early identification of rare syndromes like ED is critical for initiating appropriate supportive care and genetic counseling. We report a rare case of ED in a 1-month-old infant who initially presented with FTT and was misdiagnosed clinically as a case of progeria.

Case Report

A 1-month-old male infant was brought to our pediatric emergency unit with complaints of poor feeding, inadequate weight gain, and general lethargy. He was born at term via normal vaginal delivery with

a birth weight of 2.2 kg. There was no history of perinatal complications or significant antenatal risk factors. Family history was non-contributory.

On **clinical examination**, the infant exhibited several dysmorphic and syndromic features(Figure 1, Figure 2, Figure 3):

- **Sparse scalp hair** with a coarse, dry texture
- **Frontal bossing** with visible scalp veins
- **Deep-set eyes** with periorbital wrinkling
- **Thin, beaked nose**, and thin lips
- **Dry, wrinkled, hyperpigmented skin** particularly over the abdomen and extremities
- **Perioral and perinasal scaling and dryness**
- **Absence of eyebrows and eyelashes**
- **Hypohidrosis**, as suggested by dry skin without evidence of sweating
- **Nail dystrophy**, with thin and brittle nails
- **Generalized hypotonia** and **marked muscle wasting**
- **Prominent ribs**, sunken cheeks, and reduced subcutaneous fat stores
- **No erupted teeth** (physiological for age but significant in suspected ectodermal pathology)
- A **maculopapular hyperpigmented rash** was scattered over the limbs and torso
- Feeding was maintained via a **nasogastric (NG) tube** initially, later progressed to oral feeds

These findings contributed to an initial clinical suspicion of **Hutchinson-Gilford progeria syndrome (HGPS)** due to the aged appearance and growth failure. However, **absence of characteristic features** of progeria, such as alopecia totalis, sclerodermoid skin, joint contractures, and typical skeletal findings, led to reconsideration of the diagnosis.

Routine blood investigations including hemogram, liver and renal function tests, serum electrolytes, thyroid profile, and metabolic screening were unremarkable. Skeletal survey did not show features consistent with progeria.

In view of the atypical presentation and diagnostic uncertainty, **whole exome sequencing (WES)** was performed. The report confirmed the presence of a pathogenic variant in the *EDA* gene, confirming **hypohidrotic ectodermal dysplasia (HED)**.

The child was discharged with a high-calorie feeding plan, skin emollients, and was referred for dermatologic and genetic follow-up. Parents were provided with detailed **genetic counselling**.

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Figure 1

Figure 2

Figure 3

Discussion

Ectodermal dysplasias are typically inherited in **X-linked, autosomal dominant, or autosomal recessive** patterns [5]. Mutations in the *EDA*, *EDAR*, and *EDARADD* genes are implicated in the pathogenesis of HED [6,7]. The clinical phenotype includes varying combinations of hypotrichosis, hypohidrosis, hypodontia, nail dystrophy, and distinctive facies [1,6].

In neonates, early diagnosis can be difficult due to overlapping features with other syndromic disorders such as progeria, Seckel syndrome, and neonatal ichthyosis [8]. In our case, the **progeroid appearance**, skin features, and failure to thrive mimicked progeria,

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but molecular confirmation with **WES** enabled an accurate diagnosis.

This case highlights the **importance of clinical suspicion**, recognition of subtle features such as sparse hair, dry skin, absent eyelashes, and nail changes, and the critical role of genetic testing in atypical or syndromic FTT presentations [9].

Management of ED is **supportive and multidisciplinary**, focusing on skin care, temperature regulation, infection prevention, nutritional rehabilitation, and parental education. Long-term complications may include dental anomalies, recurrent respiratory infections, and heat intolerance [10,11].

Conclusion

This case emphasizes the need to consider **ectodermal dysplasia** in neonates presenting with failure to thrive and dysmorphic features, particularly when typical conditions like progeria are ruled out. **Whole exome sequencing** can be invaluable in resolving diagnostic dilemmas, guiding management, and enabling genetic counseling. Early recognition and supportive interventions can improve outcomes and quality of life for affected infants.

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