

Epidermolysis bullosa with col7a1 mutation at Jiangsu university hospital: A Case report and review of the literature

Fatma M Ali ¹, Jieyu Zhou ¹, Qiuxia Wang ¹, Lulu Sun ¹, Mingyan Wang ¹, Mbarouk Mohammed ² and Hongyan LUS ^{1,*}

¹ Department of Paediatrics, the affiliated hospital of Jiangsu University, Zhenjiang, Jiangsu, China.

² Department of Urology, Al-Rahma Hospital, P.O. Box 281, Zanzibar, Tanzania.

World Journal of Advanced Research and Reviews, 2023, 19(03), 451–455

Publication history: Received on 09 July 2023; revised on 18 August 2023; accepted on 21 August 2023

Article DOI: <https://doi.org/10.30574/wjarr.2023.19.3.1668>

Abstract

We describe a case of a new born Chinese male baby who was diagnosed with Epidermolysis Bullosa (EB), a rare genetic skin illness that causes particular blisters to appear on the baby's lower limbs immediately after delivery. The flaw results from the skin's reduced ability to adhere to supporting tissue, which makes it brittle. Through carrier mother inheritance, genetic testing identified our case as an EB patient with Aplasia Cutis Congenita after a nucleotide alteration in the COL7A1 gene.

Keywords: Epidermolysis Bullosa; COL7A1; Mutation

1. Introduction

A rare genetic skin condition called epidermolysis bullosa results in poor skin layer adhesion, leaving patients with thin, easily blistered skin. Both sexes are equally affected by this disorder, which typically shows up at birth or throughout infancy **(1)**. Early detection and multidisciplinary therapy can help reduce symptoms and improve outcomes even though there is no known cure **(2)**.

A variety of skin proteins, including keratins and laminin 332 type VII collagen, are impacted by the genetic abnormalities that cause EB, making the skin more brittle. Based on genetic and clinical characteristics, EB is divided into various categories. ^(1,3)

2. Case Description

We received a boy infant born in China who had skin irregularities on both of his lower limbs. Despite weighing 3.1kg and being born at full term via routine delivery without any issues, he had asymmetrical abnormalities that covered over 8% of his body surface and were mostly localized around both legs. Physical examination results revealed asymmetrical defects that were identified by thin translucent membranes with concave centres and sharp edges but no signs of bleeding or exudate. (Fig 1A), (Fig 1B)

*Corresponding author: Hongyan LUS

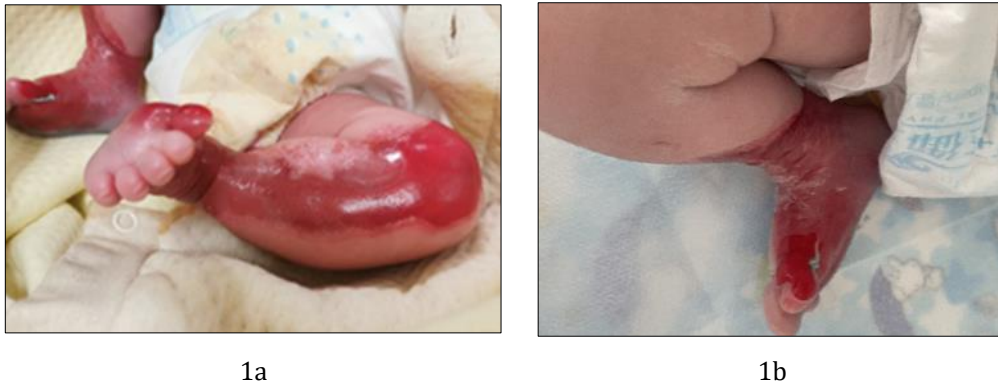


Figure 1a and 1b Skin condition of the left leg and right on the first day of birth

Following genetic testing relevant to their child's health state, which was done with both parents' informed agreement, results showed a relationship between pathogenic mutations causing an EB diagnosis and an autosomal dominant inheritance pattern. The results of the test showed that one parent had the gene variant c.6163G>A, which results in the amino acid substitution p. Gly2055Arg in the EX73/CDS73 area on the map of chromosome 3 at position 48,612,789 known for dystrophic EB compatible with the symptoms the child had. Sanger sequencing was performed to confirm the mother's status as a genetic carrier of the gene variation that caused ACC and EB development and who only had one copy of it. (Fig 2)

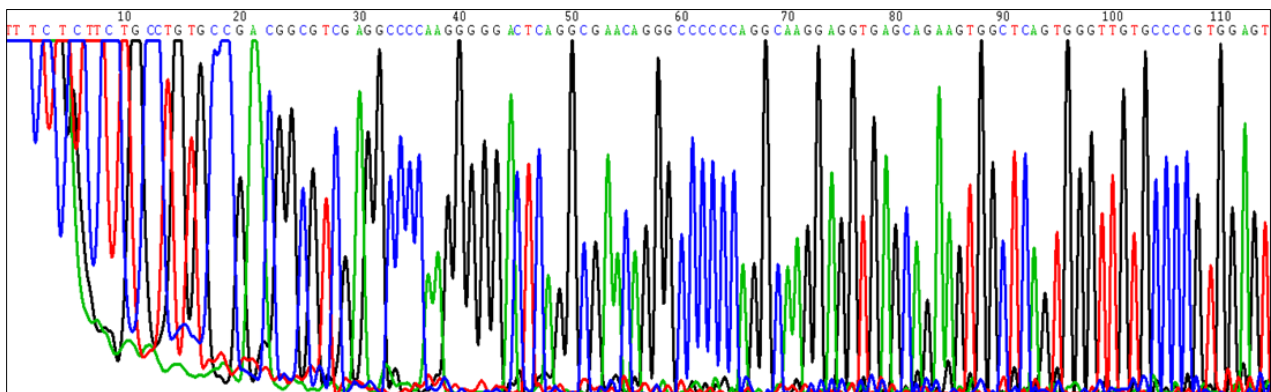


Figure 2 Sanger sequencing technology of the patient disclosing the COL7A1 gene

At the age of 32, the mother became pregnant for the first time and indicated that she was in good condition during the entire process, having never used drugs or been exposed to radiation before. The baby was stable, with a neonatal pulse of 152 beats per minute and a respiratory rate of 40 breaths per minute. Reflex tests were normal, with the exception of bilateral lower limbs. A thin, transparent membrane with a brilliant red surface covers the areas. (Fig 3A)

On the left lower extremity, there is a 15*10cm irregular "S"-shaped defect visible on the medial side of the inner lower limb. The defect extends from the medial aspect of the thumb, plantar region, medial malleolus, and the 1/2 lateral aspect of the toes, dorsum of the foot, and ankle joint, and extends to the shin of the calf up to above the knee. An unbroken skin lesion in the right calf area with a 'C' shape defect covering about 6 by 10 cm, was concave in the middle with sharp edges, and was covered by a thin film layer that made clear blood vessels apparent. (Fig 3B)

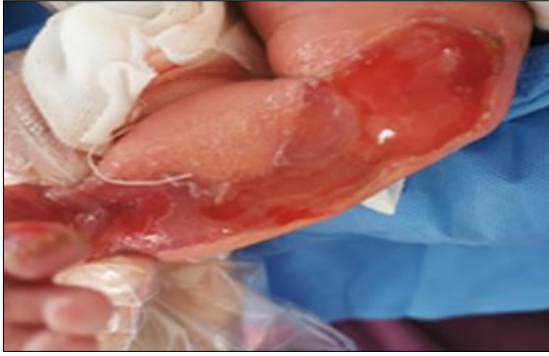


Figure 3a A thin, transparent membrane with a brilliant red surface covers the 9 areas

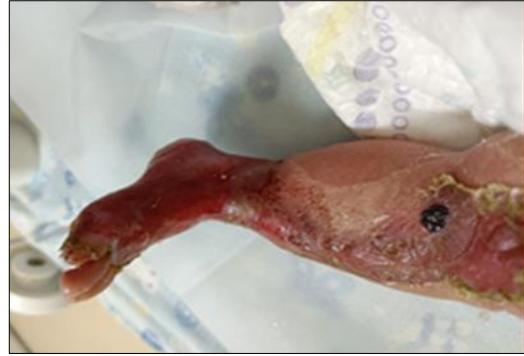


Figure 3b 'C' shape defect

A nursing session, wound assessments with sterilized water loaded with lysozyme, and polymeric foam dressing replacement every three to four days revealed a steady reduction in wound secretion until complete epithelization occurred after two days of antibiotic treatment. (Fig 4), (Fig 5)

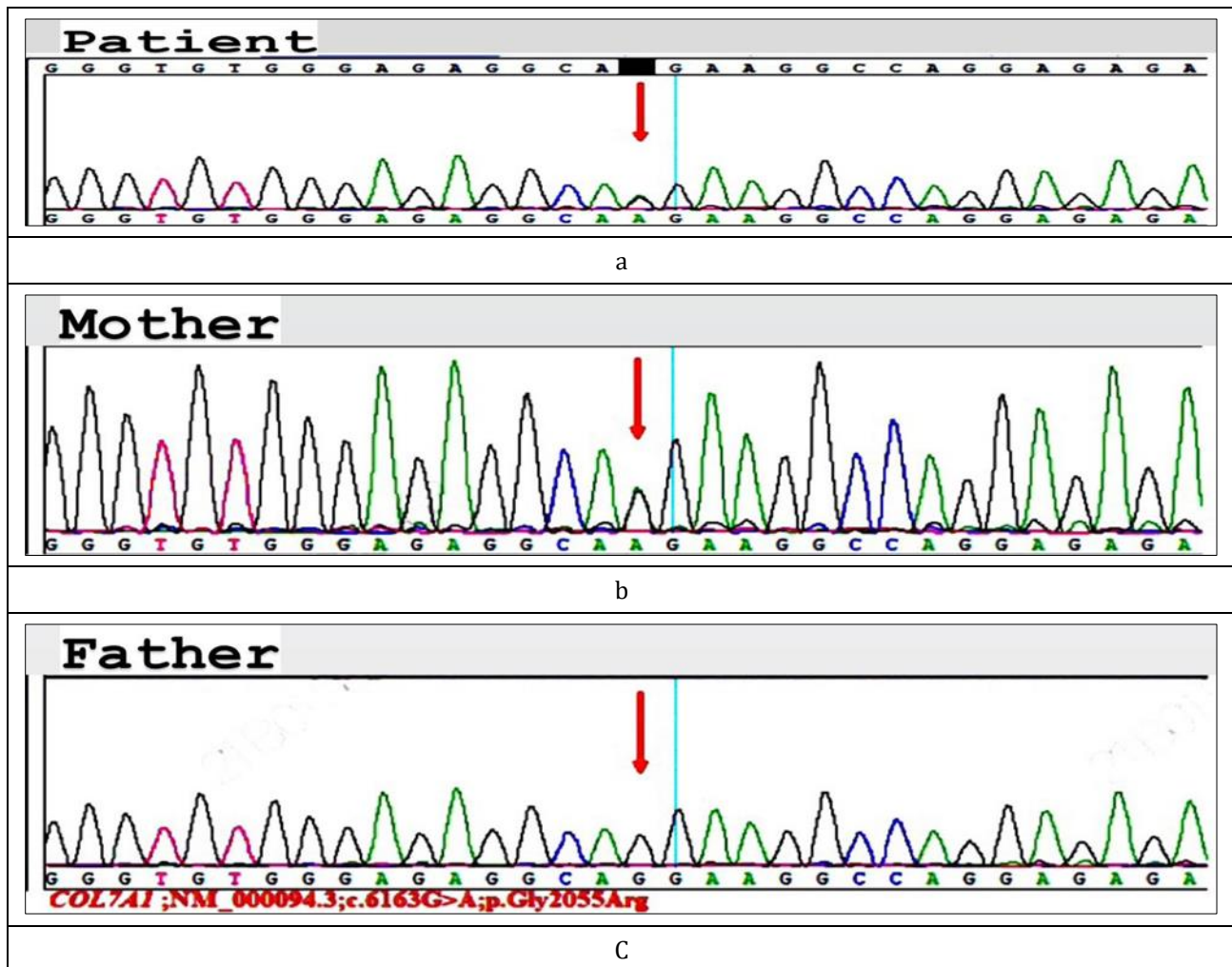


Figure 4 After wound dressing change



Figure 5 42 days after birth the 11th dressing is changed on the right leg before discharge

Although there were only a few blisters left after epithelization, the patient was able to fully recover from the anomaly affecting both lower limbs and was able to leave the hospital safely. Following a year, the infant underwent follow-up testing to look for any anomalies or growth problems. First-generation sequences from neonates with heterozygous COL7A1 gene mutations were shown. Specific blood tests, standard stool tests, and bacterial smear tests were performed. (Fig 6)



A heterozygous mutation of the affected individuals with the COL7A1(NM_000094.3) gene and a nucleotide alteration (c.6163G>A) within exon 73. An amino acid shift takes place as well (p. Gly2055Arg). This genetic variant could be found in the EX73/CDS73 region located on chromosome three at position 48,612,789. Mutated nucleotides were marked with red arrows.

Figure 6 Genetic testing results of both parents and the patient

3. Discussion

This case study article tells the story of a Chinese baby boy who was diagnosed with both EB and ACC after presenting with skin abnormalities on both lower limbs. This was accomplished based on the clinical symptoms, which were supported by the findings of the genetic testing. The COL7A1 mutation, which the child heterozygous received from the carrier mother, was discovered by genetic testing.

Prenatal gene screening was investigated by Fine et al. with positive results with the same investigation was carried out by Nishie et al to determine the relationship between recessive dystrophic EB and the underlying genetic mutations (4,5).

With the use of non-adhesive dressing changes, we managed skin fragility by preventing unnecessary damage and lowering the chance of subsequent infection with antibiotics. McCarthy and Simman et al. revealed the advantages of utilizing prophylactic antibiotics to lower the risk of infection and speed up wound healing, notably in ACC and EB patients, through the use of conservative treatment good outcomes. As evidenced by multiple instances (6,7).

4. Conclusion

GFV practitioners indeed serve a fundamental purpose in the management of these patients since it requires them to collaborate consistently for fruitful outcomes. This is done by observing the multidisciplinary approach, giving health education about the disease and its complications to the patient's next of kin, and encouraging regular clinic follow-up

Healthcare practitioners should keep in touch with the patient to keep track of any changes in the patient's developmental growth pattern and screen for any possible EB-associated complications that may arise.

Compliance with ethical standards

Disclosure of conflict of interest

All authors declare no conflicts of interest

Statement of ethical approval

This report has received approval for chart review and publication.

Statement of informed consent

Written informed consent for publication of the case details and accompanying images was obtained from the parent

Data Sharing Statement

Data to be provided upon request.

References

- [1] Gonzalez ME. Evaluation and treatment of the newborn with epidermolysis bullosa. *Semin Perinatol*. 2013 Feb 1; 37(1):32–9.
- [2] De Oliveira ZNP, Périgo AM, Fukumori LMI, Aoki V. Immunological mapping in hereditary epidermolysis bullosa. *An Bras Dermatol* [Internet]. 2010 Nov [cited 2023 May 23]; 85(6):856–61. Available from: <https://www.scielo.br/j/abd/a/P7KbQtm3SVZvCdJsHRf3HfC/?lang=en>
- [3] Krämer S, Lucas J, Gamboa F, Peñarrocha Diago M, Peñarrocha Oltra D, Guzmán-Letelier M, et al. Clinical practice guidelines: Oral health care for children and adults living with epidermolysis bullosa. *Special Care in Dentistry*. 2020 Nov 1; 40(S1):3–81.
- [4] Fine JD, Bruckner-Tuderman L, Eady RAJ, Bauer EA, Bauer JW, Has C, et al. Inherited epidermolysis bullosa: Updated recommendations on diagnosis and classification. Vol. 70, *Journal of the American Academy of Dermatology*. Mosby Inc.; 2014. p. 1103–26.
- [5] Prodinge C, Reichelt J, Bauer JW, Laimer M. Epidermolysis bullosa: Advances in research and treatment. *Exp Dermatol*. 2019 Oct; 28(10):1176–89.
- [6] McCarthy MA, Clarke T, Powell FC. Epidermolysis Bullosa and Aplasia Cutis. *Int J Dermatol* [Internet]. 1991 Jul 1 [cited 2023 May 24]; 30(7):481–4. Available from: <https://onlinelibrary.wiley.com/doi/full/10.1111/j.1365-4362.1991.tb04866.x>
- [7] Simman R. Letter to the editor: Management of aplasia cutis congenital on-scalp location (multiple letters) [1]. Vol. 57, *British Journal of Plastic Surgery*. Churchill Livingstone; 2004. p. 469–70.