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Butterfly-wing disease: is home haemodialysis possible?

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ABSTRACT

Case description: A 32-year-old woman with IgA nephropathy and "butterfly-wing disease" (dystrophic epidermolysis bullosa) required renal replacement therapy. Severe skin–mucosal fragility, even after minimal trauma, posed a major therapeutic challenge due to the difficulty of vascular access cannulation, wound care, and transfers for each haemodialysis session. Extreme care was required to prevent blistering and secondary scarring. For these reasons, a home haemodialysis technique was chosen.

Care plan description: The following nursing diagnoses were identified:

- Impaired skin integrity, manifested by bleeding and blisters, related to epidermolysis bullosa.
- Imbalanced nutrition: less than body requirements, manifested by body weight below the ideal range by age and sex and delayed wound healing, related to inadequate nutritional intake, injured oral cavity, and oesophageal stenosis.
- Chronic pain, related to cutaneous lesions caused by epidermolysis bullosa, manifested by reported fatigue and/or facial expressions of pain.
- Risk of infection.

Evaluation of the plan: Adequate training was achieved, enabling the patient to manage the technique and the identified problems, with positive changes across all outcomes established in the plan.

Conclusions: In our experience, home haemodialysis represents a viable therapeutic option in patients with dystrophic epidermolysis bullosa. It reduces hospital transfers, minimises the risk of trauma and infection, contributes to quality of life, and prolongs survival.

RESUMEN

Enfermedad en alas de mariposa: ¿es posible la hemodiálisis domiciliaria?

Descripción del caso: Mujer de 32 años, con nefropatía IgA y enfermedad de alas de mariposa (epidermólisis bullosa distrófica) que precisa terapia renal sustitutiva. La severa fragilidad cutáneo-mucosa ante mínimos traumatismos, supone un gran reto terapéutico por la dificultad de canalización de acceso vascular, las curas cutáneas y las transferencias a cada sesión de hemodiálisis. Son necesarios grandes cuidados para evitar la formación de ampollas y lesiones cicatriciales secundarias. Por todo ello, se optó por una técnica domiciliaria de hemodiálisis.

Descripción del plan de cuidados: Se identificaron los siguientes diagnósticos enfermeros:

- Deterioro de la integridad cutánea m/p sangrado y ampollas r/c epidermólisis bullosa.
- Desequilibrio nutricional: inferior a las necesidades corporales m/p peso corporal por debajo del rango ideal de peso según edad y sexo y retraso en la curación de las heridas r/c aporte nutricional inadecuado, cavidad bucal lesionada y estenosis esofágica.
- Dolor crónico r/c lesiones cutáneas causada por epidermólisis bullosa m/p expresa fatiga y/o expresión facial de dolor.
- Riesgo de infección.

Evaluación del plan: Se logró realizar un adecuado entrenamiento consiguiendo el manejo de la técnica y de los problemas detectados, con un cambio positivo en todos los NOC establecidos en el plan.

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Conclusiones: En nuestra experiencia, la hemodiálisis domiciliaria supone una opción terapéutica viable en pacientes con epidermólisis bullosa distrófica. De esta manera, se disminuye el número de traslados al hospital, minimizando el riesgo de traumatismos e infecciones, contribuyendo a la calidad de vida del paciente y prolongando su supervivencia.

INTRODUCTION

Dystrophic epidermolysis bullosa (DEB), also known as "butterfly skin disease," is a rare genetic disorder caused by mutations in the COL7A1 gene, responsible for the synthesis of type VII collagen (C7), an essential protein for dermo-epidermal adhesion. Its recessive form (RDEB) is characterized by a marked reduction or absence of C7, leading to extreme fragility of the skin and mucous membranes, spontaneous blister formation or blistering after minimal trauma, and resulting in skin contractures, digital fusions, and severe gastrointestinal strictures[1]. As wounds heal, the skin loses elasticity and joint mobility becomes progressively limited, compromising patient autonomy.

The estimated prevalence is 2 cases per 100,000 people, according to the Dystrophic Epidermolysis Bullosa Research Association (DEBRA) (DEBRA Butterfly Skin Association, 2025). Despite its low frequency, DEB entails a high burden of morbidity and a reduced life expectancy, mainly conditioned by infectious complications, cutaneous neoplasms, and severe malnutrition.

The association between DEB and chronic kidney disease (CKD) has been scarcely documented in the literature. The first reported case of renal involvement dates back to 1973 and was attributed to secondary genitourinary complications[2]. Subsequently, isolated cases of glomerulopathies have been reported, including IgA nephropathy, amyloidosis, or glomerulonephritis, as in the case presented here[3].

The coexistence of DEB and advanced CKD constitutes an exceptional and highly complex clinical situation. Extreme cutaneous–mucosal fragility represents a significant barrier to conventional renal replacement therapy, complicating vascular access, routine wound care, and frequent hospital transfers. In addition, compromised nutritional status and a high risk of infection further worsen clinical outcomes.

In this context, home hemodialysis emerges as an optimal, safe, and personalized therapeutic alternative capable of minimizing risks associated with the hospital environment and improving patient quality of life. The present case report provides a relevant contribution to the scarce existing evidence and offers practical insight into the comprehensive management of patients with rare diseases and complex nephropathies from a multidisciplinary and individualized perspective.

CASE PRESENTATION

A 32-year-old woman with recessive dystrophic epidermolysis bullosa since birth. In this context, the patient presents episodic chronic dysphagia due to esophageal stenosis, requiring periodic dilations, and has had a permanent gastrostomy since age three. She required extraction of six teeth and two oral fibromas (dental prosthesis pending evaluation). She has syndactyly and multiple squamous cell carcinomas that required surgical excision. She has had numerous episodes of cutaneous and urinary tract infections.

From a renal perspective, she has advanced chronic kidney disease secondary to diffuse mesangial IgA nephropathy. Due to her unique clinical circumstances—living 80 km from the hospital, requiring multiple visits to her reference center and to other specialists—a home-based renal replacement therapy was selected. Because she has had a permanent gastrostomy since childhood, she had a relative contraindication for peritoneal dialysis, and home hemodialysis was chosen.

Upon initial evaluation, several active problems were identified, including protein–calorie malnutrition, with a body mass index of 16.52% (due to chewing difficulty, low intake, and a history of refeeding syndrome), as well as difficulty establishing vascular access, with absence of local anesthetic effect.

Hemodialysis initiation was scheduled, and a tunneled pediatric right internal jugular central venous catheter was placed. The modality was agreed upon with the patient and her family, prioritizing quality of life and minimizing hospital visits to allow continuation of her daily wound-care protocols (every 12 hours, lasting approximately 2 hours) and reduce infection risk.

Training took place in the hospital and included two family members in a semi-assisted educational phase (family/partner present and directed by the patient). Training lasted 21 days (150-minute sessions), three days per week, due to the patient's physical fatigue. In parallel with the medical team's training of the family, we also provided comprehensive education—with support from the DEBRA association—regarding the care required for the disease. Given the risk of injury in DEB, all routine clinical procedures were adapted to the patient's complexity: blood pressure was measured using devices without direct compression; soft silicone dressings were used to avoid damage during removal; bed sheets were kept wrinkle-free to reduce friction; identification was made with cards at the bedside or labels on clothing, avoiding wristbands. Monitoring used special soft-adhesion electrodes and sensors protected with non-adherent dressings to minimize any risk of skin injury.

During 2 years on the technique, monthly evaluations were scheduled, and the patient required multidisciplinary follow-up. She was hospitalized four times: once for heart failure, once for infectious diarrhea (Campylobacter), and

twice for bacteremia. She required surgical removal of multiple cutaneous lesions (squamous cell carcinoma). Major complications included: absence of anesthetic effect for surgical procedures, catheter dysfunction, difficult pain management, limited vascular access, rapid loss of residual renal function, high nutritional requirements, anemia management, and anxiety–depression.

Her most recent hospital admission was due to septic shock secondary to *Staphylococcus aureus* bacteremia and COVID infection. Severe coughing episodes caused oral blisters and ulcers, making oral intake impossible. During the admission, transfers to the hemodialysis unit (skin friction) produced extensive lesions on the back with significant disruption of the skin barrier, blisters, and retractile scarring (over 80% body surface area).

Despite initially appearing impossible to perform hemodialysis in this patient, after more than a year and a half on renal replacement therapy, she remains stable on home hemodialysis.

COMPREHENSIVE NURSING ASSESSMENT ACCORDING TO THE MARJORY GORDON MODEL

To conduct the nursing evaluation, the "Assessment Guide for Patients with End-Stage Chronic Kidney Disease on Hemodialysis"[4] was used. This document incorporates tools such as NANDA taxonomy and the Nursing Outcomes Classification (NOC), and is based on Marjory Gordon's functional health patterns model[5]. Its purpose is to promote the use of a common nursing language and support comprehensive, standardized care.

Below is the assessment according to the 11 functional health patterns, including both altered and preserved aspects, consistent with the model's holistic approach.

Pattern 1 – Health perception and health management
The patient is knowledgeable about her condition, actively involved in her own care, adherent to treatment, and participates in decision-making.

Pattern 2 – Nutritional–metabolic
Protein–calorie malnutrition (Body Mass Index of 17) associated with epidermolysis bullosa, with oral blisters, missing teeth, esophageal stenosis, high insensible losses, and hydroelectrolytic imbalances (hypokalemia and hypophosphatemia). She maintains fluid restriction due to anuria.

Pattern 3 – Elimination
Persistent anuria due to chronic kidney disease. Altered bowel elimination: frequent diarrheal episodes related to recurrent digestive infections and chronic inflammatory status.

Pattern 4 – Activity and exercise
High dependency for basic activities of daily living. Very limited mobility due to joint contractures and skin lesions. The patient remains in a supine position most of the time and requires support devices and assisted mobilization. Reports habitual fatigue.

Pattern 5 – Sleep and rest
Fragmented, non-restorative sleep. Environmental and pharmacological interventions are required to improve rest.

Pattern 6 – Cognitive–perceptual
Alert and oriented in all three spheres. No cognitive impairment. Reports persistent, difficult-to-control chronic pain due to blistering lesions, with poor response to local anesthetic agents.

Pattern 7 – Self-Perception and self-concept
Presents altered body image, feelings of frustration, and dependency. Despite this, she maintains a cooperative attitude and active coping.

Pattern 8 – Role and relationships
Highly dependent on her environment. Primary caregivers are significantly involved in care. Family relationships are functional and provide strong emotional support.

Pattern 9 – Sexuality and reproduction
The patient has a stable partner. The sexual domain has not been explored in depth, as she did not express concerns regarding this pattern during the assessment. No current needs were identified.

Pattern 10 – Coping and stress tolerance
Demonstrates good adaptive capacity toward her chronic disease. Uses active coping strategies and benefits from effective emotional support from her family. Upon the need to initiate renal replacement therapy, she displayed signs of anxiety and emotional distress.

Pattern 11 – Values and beliefs
No ethical or spiritual conflicts identified. She respects therapeutic indications and actively participates in treatment-related decisions.

EVALUATION OF THE CARE PLAN

Nursing intervention outcomes were assessed using NOC indicators, with partially favorable clinical progress observed across several domains. Improvements were notable in nutritional status, pain control, and adherence to fluid management, although complications related to extreme cutaneous–mucosal fragility and chronic malnutrition persisted.

Nutritional management
Specific oral supplementation was initiated, along with parenteral nutrition during hemodialysis sessions. Nutritional

Description of the NANDA–NIC–NOC care plan.

Nursing Diagnosis (NANDA)	NOC (Outcome / Indicator)	NIC (Nursing Intervention)
(00002) Imbalanced nutrition: Less than body requirements, related to DEB-associated conditions.	(1004) Nutritional status.	(1100) Nutrition management.
	(100402) Food intake.	(1260) Weight management.
	(1260) Weight management.	(5614) Teaching: Prescribed diet.
		(1200) Parenteral Nutrition administration during hemodialysis.
(00195) Risk for electrolyte imbalance, related to renal dysfunction.	(0601) Fluid balance.	(4120) Fluid management.
	(1092) Risk control.	
	(060112) Absence of peripheral edema.	
	(190802) Acknowledges risk.	
	(190207) Uses risk-control strategies.	
(00095) Insomnia, related to inadequate sleep hygiene, manifested by non-restorative sleep.	(0004) Sleep.	(1850) Improve sleep.
	(000402) Hours of sleep.	
	(000404) Sleep quality.	
(00094) Risk for activity intolerance, related to imbalance between oxygen supply and demand.	(1804) Energy conservation.	(0180) Energy management.
	(180403) Appropriate activities.	
(00004) Risk for Infection, manifested by chronic DEB involvement and/or presence of central venous catheter, related to impaired skin integrity.	(1908) Risk detection.	(6540) Infection control.
	(190801) Recognizes signs and symptoms of risk.	(3590) Skin surveillance.
	(3102) Self-management: chronic disease.	(1800) Assistance with self-care.
(00002) Risk for falls, related to physiological factors.	(1828) Knowledge: fall prevention.	(6490) Fall prevention.
	(182801) Description of use of assistive devices.	(6486) Environmental management: safety.
	(182817) Description of How to Ambulate Safely.	
(00046) Impaired skin integrity, related to DEB and AVF, manifested by skin lesions.	(1102) Wound healing: primary intention.	(3590) Skin surveillance.
	(1105) Hemodialysis access integrity.	(3660) Wound care.
	(110303) Purulent drainage.	(3440) Incision Site care.
	(110322) Wound inflammation.	
	(110311) Skin blisters.	
(00045) Impaired oral mucous membrane, related to mechanical factors (pressure, friction, shear), manifested by lesions.	(1100) Oral hygiene.	(1730) Restoration of oral health.
	(110012) Oral mucous membrane integrity.	(3590) Skin surveillance.
(00133) Chronic pain, related to epidermolysis bullosa lesions, manifested by verbal reports of pain.	(1605) Pain control.	(1400) Pain management.
	(160511) Reports pain controlled.	(6040) Simple relaxation therapy.

DEB: Dystrophic epidermolysis bullosa.

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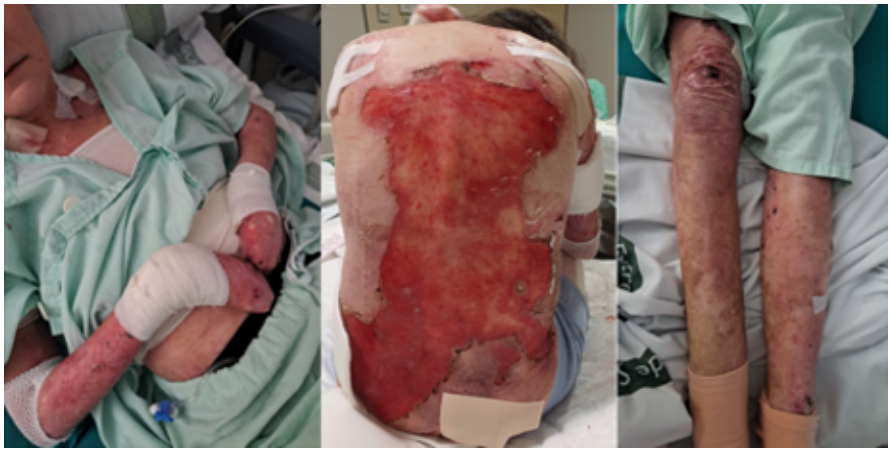

Image 1. Severe skin involvement due to friction during an intrahospital transfer.

education was provided and nutritional status was monitored via bioimpedance. The patient initially had a BMI of 16.5, which increased slightly to 17.1 over three months. Estimated daily caloric intake increased from <1000 kcal to approximately 1050 kcal, representing a limited yet sustained improvement. Electrolyte disturbances associated with malnutrition, such as hypophosphatemia and hypokalemia, were corrected through oral and parenteral supplementation, with progressive laboratory normalization. Improved NOC indicators: *Nutritional Status* (1004) and *Food Intake* (100402).

Fluid management

Continuous monitoring of fluid balance was performed. Dry weight remained stable (36.2 kg to 36.0 kg). No signs of fluid overload, edema, or hypotensive episodes were observed. Adherence to fluid restriction recommendations was good, and electrolyte stability was maintained after correction of initial hypokalemia. NOC indicators: *Fluid Balance* (0601) and *Absence of Peripheral Edema* (060112).

Pain management

Chronic pain, initially rated 8/10 on a visual analog scale, decreased to 5/10 following optimization of pharmacological therapy and coordination with the pain unit. Relaxation techniques and non-pharmacological measures were added. Control was partial, with pain exacerbations during mobilization or infections. NOC indicators: *Pain Control* (1605) and *Reports Controlled Pain* (160511).

Trauma prevention

Caregivers were trained in safe mobilization techniques, and the home environment was adapted (special mattresses, wrinkle-free sheets, non-adherent dressings). No falls occurred at home within the first three months; however, during a subsequent hospital admission, an improper transfer resulted in extensive skin injury **(image 1)**. Thus, although traumatic episodes decreased at home, risk remains high in uncontrolled settings. Partial improvement was observed in: *Fall Prevention* (NOC 6490), and *Skin Surveillance* (NOC 3590)

DISCUSIÓN

The coexistence of recessive dystrophic epidermolysis bullosa (RDEB) and advanced CKD represents a significant clinical challenge due to its low prevalence, therapeutic complexity, and high morbidity burden. Although rarely described, renal involvement in patients with RDEB is not anecdotal. In a multicenter study conducted by the National Epidermolysis Bullosa Registry, up to 12.3% of patients with Hallopeau–Siemens–type RDEB died from renal causes before the age of 35, establishing renal failure as one of the main causes of death in this young population[6].

Renal complications described in this population include primarily IgA nephropathy, secondary amyloidosis, and post-infectious glomerulonephritis[7,8]. These conditions may progress silently; therefore, regular monitoring of renal function from early stages is recommended. This aspect is particularly relevant in patients with recurrent skin infections, chronic inflammatory states, and malnutrition—factors commonly present in the evolution of RDEB.

Interventions and NOC Indicators.

Intervention	NOC Indicator	Initial Value	Final Value	Estimated Improvement
Nutritional Management	BMI (kg/m²)	16.5	17.1	+3.6%
	Daily caloric intake (100402)	<1000 kcal	~1050 kcal	+5–10%
	Phosphatemia, potassium levels	Altered	Normalized	100% resolved
Fluid Management	Peripheral edema (060112)	Present (mild)	Absent	100% resolved
	Fluid balance	Variable	Stable	100% controlled
Pain Management	VAS pain scale	8/10	5/10	–37.5%
Trauma Prevention	Friction injuries	Frequent	Sporadic	>50% reduction
	Falls (at home)	1 episode per month	0 (in 3 months)	100% in controlled environment

Renal replacement therapy in patients with DEB presents multiple challenges due to their particular cutaneous–mucosal fragility. Successful cases of hemodialysis, peritoneal dialysis, and even kidney transplantation have been reported, always with adaptations tailored to patient characteristics[9,10]. Peritoneal dialysis, despite its apparent lower aggressiveness, may be contraindicated in the presence of abdominal involvement, high risk of peritonitis, or prior gastrostomies. Consistent with published data, in our case this modality was ruled out due to the presence of a permanent gastrostomy and the associated technical risk[11]. Hospital-based hemodialysis carries risks related to frequent manipulation of vascular access, pressure from medical devices on the skin, and recurrent transfers[12,13]. In our experience, even minimal mobilizations triggered extensive lesions on the patient's back and oral mucosa, clearly illustrating the difficulty of maintaining this modality without worsening her overall condition. Similar complications have been reported by Małecki et al., who emphasized the need to rigorously adapt dialysis care to avoid skin injury in these patients[9].

Given these circumstances, home hemodialysis emerges as an optimal therapeutic alternative. By eliminating the need for transportation, allowing careful handling in a controlled home environment, and adapting techniques and devices to individual needs, this modality becomes a safe and effective option to preserve skin integrity and reduce complications[14]. In our case, the technique was successfully maintained for more than 18 months through a progressive training program, family involvement, and close clinical supervision.

Finally, although kidney transplantation remains a valid alternative, its application in patients with RDEB requires meticulous evaluation. Successful cases have been reported following specific surgical and anesthetic preparation, suggesting that with proper planning, transplantation may be considered in advanced stages, particularly in young patients with limited extra-renal comorbidity[11].

Overall, this clinical case adds valuable evidence regarding the feasibility of home hemodialysis in patients with RDEB and advanced CKD. Management requires a multidisciplinary approach, individualized protocols, and strong commitment from both the family environment and healthcare professionals. The accumulated experience, combined with existing literature, reinforces the need to disseminate such cases to guide future therapeutic decisions in rare diseases of high complexity.

CONCLUSIONS

This clinical case demonstrates that even in highly complex clinical scenarios—such as a patient with dystrophic epidermolysis bullosa and advanced chronic kidney disease—it is possible to successfully implement home hemodialysis when an individualized care plan, adequate training, and continuous support are provided. Over the course of follow-up, objective clinical improvements were observed in indicators such as nutritional status, pain control, and prevention of skin injuries, confirming that the goals established in the care plan were effectively achieved.

The patient's favorable evolution reinforces the idea that complex therapeutic options should not be dismissed solely due to their technical difficulty. This case highlights the importance of evaluating each situation comprehensively and with flexibility, tailoring available resources and strategies to the patient's specific characteristics, even when physical fragility and environmental conditions pose constant challenges. Nursing intervention—focused on accompaniment, environmental adaptation, and caregiver education—was key to ensuring treatment safety and continuity.

The strong involvement of all healthcare personnel, the support of the DEBRA Association in Spain, and the emotional and physical commitment of the patient's family were fundamental pillars in achieving effective coping during this new stage of life.

Our experience shows that home hemodialysis is not only feasible in patients with epidermolysis bullosa, but may also represent an alternative that improves quality of life, preserves autonomy, and dignifies the therapeutic process.

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Conflicts of interest

None declared.

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