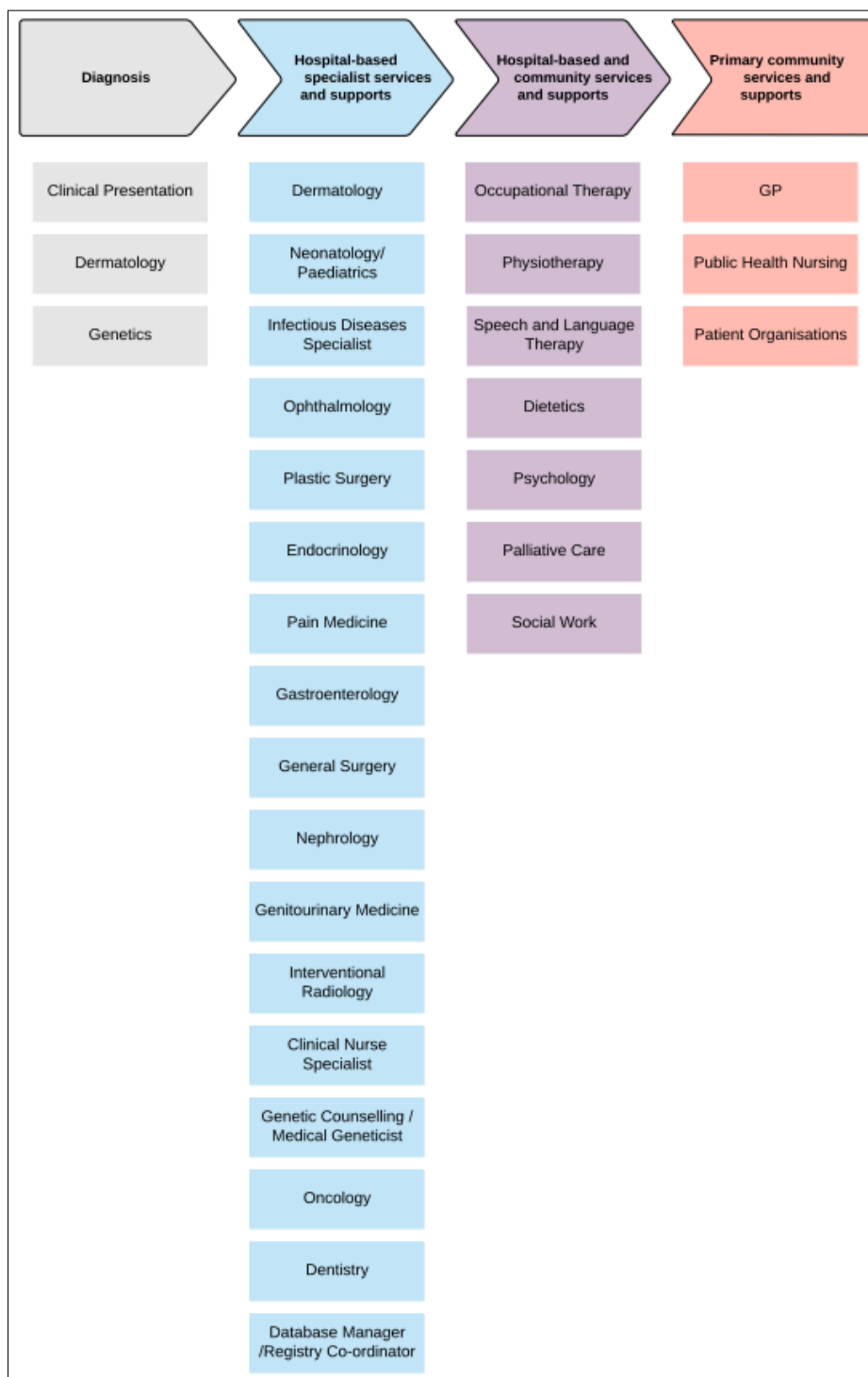




Inherited Epidermolysis Bullosa (EB)

Paediatric & Adult Care Pathway

Inherited Epidermolysis Bullosa (EB) Paediatric & Adult Care Pathway



Clinical characteristics:

Orphacode: 79361

Disease definition: Inherited epidermolysis bullosa (EB) encompasses a number of disorders characterized by recurrent blister formation as the result of structural fragility within the skin and selected other tissues

Diagnosis	Clinical Presentation	Clinically variable ranging from localized blistering of the hands and feet to generalized blistering of the skin, oral cavity, and mucous membranes with multi-organ involvement; milia; nail hypoplasia; nail dystrophy; alopecia; abnormality of skin pigmentation
Diagnosis	Dermatology	Diagnosis and classification of EB type and subtype is based on the level of cleavage at the dermal/epidermal junction - clinical examination, detailed personal and family history, immunofluorescence antigen mapping and / or transmission electron microscopy of skin biopsy
Diagnosis	Genetics	Next Generation Sequencing multigene panel
Hospital-based specialist services and supports	Dermatology	Management of skin fragility and wound infections. Management of associated coexisting issues – nutrition, growth, pain, pruritis, and mobility. Screening for skin malignancy. Enrolment in clinical trials. Rare Disease Registry – National EB Registry operating through National and International Skin Registry (NISR charity)
Hospital-based specialist services and supports	Neonatology/ Paediatrics	Support during neonatal period, management of general health and of acute episodes of illness
Hospital-based specialist services and supports	Infectious Diseases Specialist	Management of complicated infections
Hospital-based specialist services and supports	Ophthalmology	Corneal erosion, corneal scarring
Hospital-based specialist services and supports	Plastic Surgery	Release of contractures, Hand Reconstruction, surgical management of skin cancers
Hospital-based specialist services and supports	Endocrinology	Management of delayed puberty. Optimising bone health. Management of osteopenia/ osteoporosis
Hospital-based specialist services and supports	Pain Medicine	Management of acute and chronic pain and pruritis
Hospital-based specialist services and supports	Gastroenterology	Investigation and management of enteropathy and chronic constipation
Hospital-based specialist	General Surgery	Gastrostomy insertion, management of pyloric stenosis

services and supports		
Hospital-based specialist services and supports	Nephrology	Screening and management of kidney disease and renal failure
Hospital-based specialist services and supports	Genitourinary Medicine	Screening and management for genitourinary malformations
Hospital-based specialist services and supports	Interventional Radiology	Imaging and management of oesophageal stenosis (barium studies, videofluoroscopy, balloon dilatation)
Hospital-based specialist services and supports	Clinical Nurse Specialist	Education and support for patients and carers Skin care and wound management Training and support of home care teams Support during periods of transition in education and employment Coordination of multidisciplinary care
Hospital-based specialist services and supports	Genetic Counselling / Medical Geneticist	Genetic counselling, recurrence rates, reproductive options
Hospital-based specialist services and supports	Oncology	Medical management of skin cancers
Hospital-based specialist services and supports	Dentistry	Yearly review for caries, dental hypoplasia, oral health
Hospital-based specialist services and supports	Database Manager/Registry Co-ordinator	Create and maintain database of patients attending service Record patient biographical, clinical and research data Audit and quality improvement Ensure minimum data set standards for ERN registry National EB Registry operating through National and International Skin Registry (NISR charity)
Hospital-based and community services and supports	Occupational Therapy	Assessment and intervention for difficulties participating in activities of daily living; including play, sleep, self-care, fine and gross motor skills. This may include environmental adaptations, aids and appliances to promote independence. Upper limb assessment and intervention. This may include hand orthoses and home exercise programme to preserve hand function and maintain ROM and promote independence Postural management assessment and intervention, including specialist seating and equipment provision Feeding and mealtime support

		<p>Home and school environmental assessments and adaptation recommendations</p> <p>Assessment for handwriting and assistive technology for school</p> <p>Sensory processing assessment and intervention</p>
Hospital-based and community services and supports	Physiotherapy	<p>Neuromusculoskeletal: Skin fragility, Scarring, fibrosis (webbing) and contractures affecting any body part, fatigue and decreased endurance, strength & balance deficits, pain and pruritus, gross motor skill delay</p> <p>Educate on avoidance of secondary injury by purposefully handling body and limbs strategically – delay contractures and deformities</p> <p>Identify range of movement limitations, muscle power limits, skin integrity and trigger spots of friction to assess need for socks, orthotics or shoes</p> <p>Bandage wrapping techniques</p> <p>Facilitate attainment of developmental motor milestones through developmental activities and positioning</p> <p>Enhance bone health, functional activities and endurance training through safe weight-bearing, stretching, strengthening and balance exercises</p> <p>Maintenance and support programs to prevent recurrence, re-injury or functional decline – enhance patient autonomy</p> <p>Provide mobility devices</p> <p>Rehabilitation post-surgical correction</p> <p>Pain management strategies</p> <p>Avoid use of adhesive tapes, dressings and felt</p> <p>Tricycles and mobility scooters have been recommended as an alternative to wheelchairs for this patient group</p>
Hospital-based and community services and supports	Speech and Language Therapy	<p>Assessment, support and intervention for feeding, eating, drinking & swallowing (FEDS) skills based on individual needs and priorities</p> <p>Advocacy, education and training of family, staff, educators to support patient’s participation, activities, learning, independence and social-emotional well-being across their environments</p>
Hospital-based and	Dietetics	<p>Nutrition assessment including monitoring of growth and regular screening for malnutrition to ensure adequate nutrition intake</p>

community services and supports		<p>Monitoring and provision of nutritional support including enteral tube feeding</p> <p>Ongoing monitoring and management of iron deficiency anaemia, chronic micronutrient deficiency and constipation</p> <p>Dietary advice to optimise bone health and wound healing</p> <p>Dietary management of renal impairment</p> <p>Dietary advice on provision of texture modified diets where aversive oral feeding behaviour or swallowing difficulties arise</p> <p>Management of dysphagia secondary to oesophageal strictures</p>
Hospital-based and community services and supports	Psychology	Support for chronic condition, anxiety, depression, coping strategies for patients and carers
Hospital-based and community services and supports	Palliative Care	Pain and symptom relief End of life care
Hospital-based and community services and supports		<p>Psychosocial support: Assess social and family supports, safeguarding</p> <p>Link with community supports as required e.g., GP, Public Health Nurse, Primary Care SW, Local authority SW, Mental Health SW, Disability SW, TUSLA and DEBRA Ireland</p> <p>Offer counselling or GP referral to Counselling in primary care (CIPC) www.hse.ie/eng/services/list/4/mental-health-services/counsellingpc/</p> <p>Financial support (as required): Patient advocacy, support applications for Medical card, Disability allowance, Supplementary Welfare allowance, Exceptional Needs payment, Long-term illness card, direct to Citizens' information www.citizensinformation.ie/</p> <p>Housing and/or mobility issues: Advocacy and support</p> <p>Home Care Packages: if issues with activities of daily living - arrange application for inpatients, liaise with Public Health Nurse to arrange for out-patients</p> <p>Respite Care: liaise with public health nurse or community disability services to arrange</p> <p>Employment issues: Link to Intreo public employment and EmployAbility services</p>

<p>www.gov.ie/en/campaigns/fb84c0-intreo/ www.gov.ie/en/service/8578c4-access-the-employability-service/</p>		
Primary/ community services and supports	GP	Co-ordinator of community-based supports Management of general health Key link with secondary/ tertiary care
Primary/ community services and supports	Public Health Nursing	Home care nursing to support dressing changes School support e.g. SNA
Primary/ community services and supports	Patient organisations	Advocacy, support and information: DEBRA Ireland (www.debraireland.org) - Counselling and Therapy Grant (funds counselling and play therapy sessions) and peer-to-peer supports ERN Skin (www.ern-skin.eu) EB Clinet (www.eb-clinet.org) Rare Ireland Family Support Network (www.rareireland.ie) Rare Diseases Ireland (www.rdi.ie)

Clinical Lead:

Dr. Fiona Browne, CHI at Temple Street and St. James' University Hospital, Dublin

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