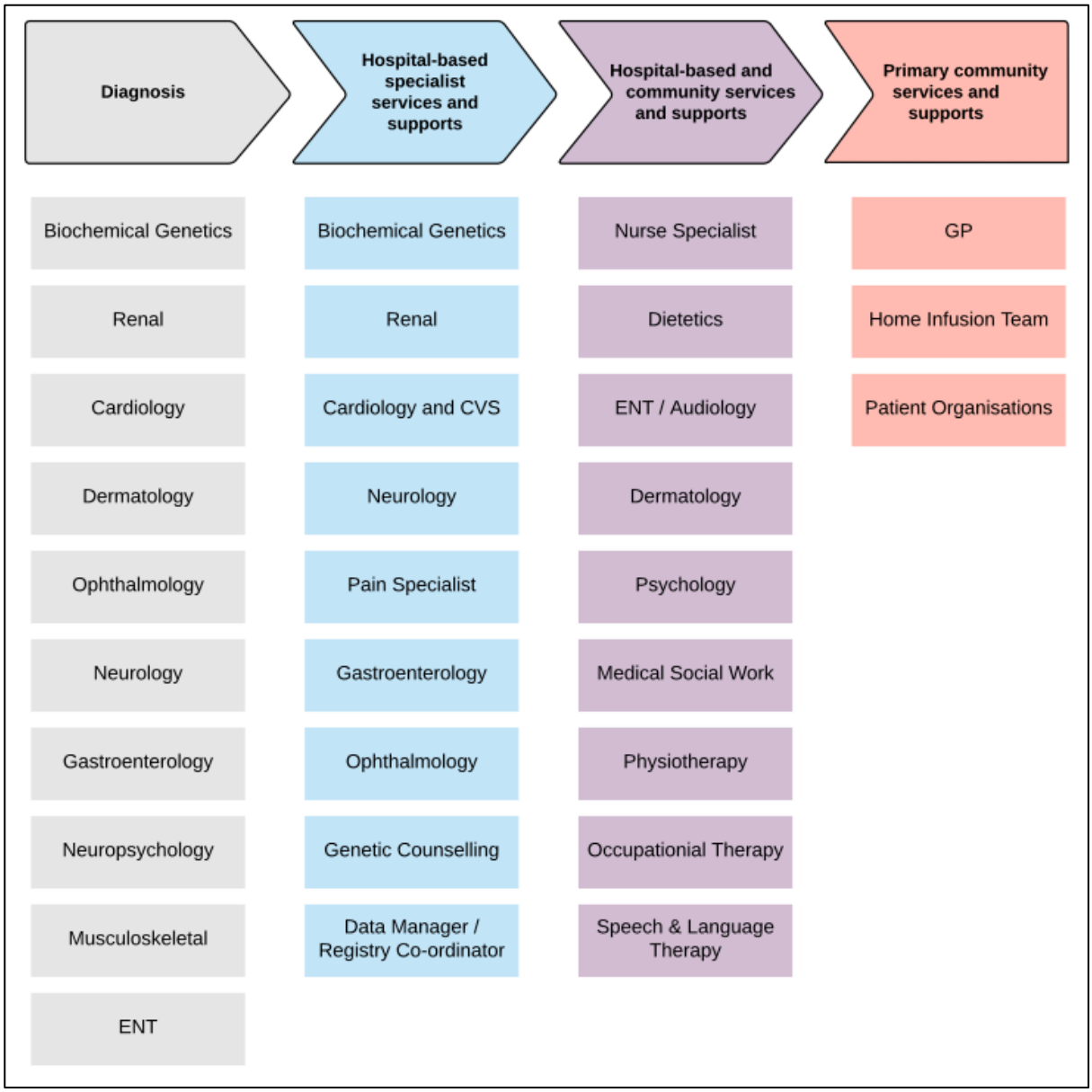
Fabry Disease

Adult Care Pathway

HSE National Rare Diseases Office

Fabry Disease (FD) Adult Care Pathway



Clinical Characteristics:

Orphacode: **324**

Disease Definition: Fabry Disease (FD) is a progressive, inherited, multisystemic lysosomal storage disease characterised by specific neurological, cutaneous, renal, cardiovascular, cochlea-vestibular and cerebrovascular manifestations

Diagnosis	Biochemical Genetics	Known pathogenic variant in *GLA* gene; Alpha-gal activity in males; Gene analysis and enzyme in females; Increased plasma and/ or urinary GL-3 or plasma lyso GL-3; Angiokeratoma, cornea verticillata, renal biopsy, facial gestalt, acroparesthesia
Diagnosis	Renal	Unexplained decreased GFR/ Kidney failure; Pathological albuminuria/ proteinuria; Renal Biopsy; Hypertension
Diagnosis	Cardiology	Unexplained cardiomyopathy (particularly hypertrophic with concentric hypertrophy); Reduced exercise tolerance; Syncope; Cardiac fibrosis; Heart Failure; Atrial fibrillation or conduction defects; Valvular disease; Dilation of aorta; Dyslipidaemia
Diagnosis	Dermatology	Angiokeratomas, hypohydrosis/ hyperhidrosis, lymphoedema
Diagnosis	Ophthalmology	Cornea verticillata; Rare central retinal artery occlusion; Cataract
Diagnosis	Neurology	Neuropathic pain; Acroparesthesia; Transient ischaemic attacks (TIAs); Strokes; Cerebral venous thrombosis/ Hypertension
Diagnosis	Gastroenterology	Nausea; Vomiting; Diarrhoea; Constipation; Abdominal pain/ bloating
Diagnosis	Neuropsychology	Common – depression, anxiety, panic attacks; Cognitive decline and dementia (rare)
Diagnosis	Musculoskeletal	Osteopenia; Osteoporosis
Diagnosis	ENT	Sensorineural hearing loss, Tinnitus, Vertigo
Hospital-based specialist services and supports	Biochemical Genetics	Provide copy of Genetic report to all patients Treatment - Enzyme replacement therapy or pharmacological chaperone (Galafold) according to guideline for initiation see (Ortiz et al, 2018) Monitoring biomarkers, Plasma Lyso GB3 and urinary GB3, identification of other family members at risk

		Yearly review – clinical exam including plasma and urine biomarkers Screen for osteopenia & osteoporosis, BMD every two years, if unremarkable, Refer to Endocrinology, if required Refer Audiology, ENT if required ECG, echo and refer Cardiology Stroke prophylaxis: anticoagulants (if indicated by history) Future - possible Substrate Reduction Therapy and Gene therapy Clinical trials Establish links with regional centres to develop shared care where possible – accommodate regional blood testing prior to specialist clinic visits
Hospital-based specialist services and supports	Renal	ACE Inhibitor (ACEI) or angiotensin-II receptor blockers (ARB) to target albuminuria and proteinuria Management of CKD, GFR annually, every 3 months if high risk, Statin administration Monitor vitamin D status Refer to dietitian for dietary management of CKD and associated complications (KDOQI, 2020) Dialysis or kidney transplantation for patients entering renal failure
Hospital-based specialist services and supports	Cardiology and CVS	ACEI or ARBs; ECG and echo with strain-annually; Cardiac MRI for cardiomyopathy (prn) with gadolinium enhancement and T1 mapping (if available); Blood pressure surveillance; 48-hour holter monitor; Manage arrhythmias/ cardiac pacing; Anticoagulation for arrhythmias, implantable cardioverter-defibrillator (ICD) placement; Biomarkers - BNP or NT BNP pro
Hospital-based specialist services and supports	Neurology	Brain MRI (if symptomatic)
Hospital-based specialist services and supports	Pain Specialist	Pain management in specialist centre if indicated or regional hospital (as required) - Neuropathic pain, headache
Hospital-based specialist services and supports	Gastroenterology	Metoclopramide for delayed gastric emptying Refer to dietitian for dietary interventions
Hospital-based specialist services and supports	Ophthalmology	Polarized glasses Artificial tears
Hospital-based specialist	Genetic Counselling	Review X-linked inheritance, recurrence risk, reproductive options, identify at-risk relatives, cascade testing

services and supports		
Hospital-based specialist services and supports	Data Manager / Registry Co-ordinator	Create & 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
Hospital-based and community services and supports	Nurse Specialist	Patient and family education, advice, and support Liaise with health care professionals to accomplish holistic personal care pathways in hospital and community Main point of contact for patients Link to patient organisations
Hospital-based and community services and supports	Dietetics	Gastroenterology: Metoclopramide for delayed gastric emptying, H-2 blockers for dyspepsia symptoms Dietary interventions for dysmotility and diarrhoea FODMAP diet consideration Monitor growth in paediatrics as gastrointestinal symptoms may affect overall intake Nutrition assessment and nutrition support for people identified with delayed growth, failure to thrive or malnutrition Renal: Monitor vitamin D status and replacement therapy if deficient. Dietary management of chronic kidney disease (CKD) and associated complications CKD Mineral Bone Disorder prevention and management
Hospital-based and community services and supports	ENT/ Audiology	Hearing aids; Cochlear implants; Audiology review 2 yearly (if normal), if abnormal (as required)
Hospital-based and community services and supports	Dermatology	Laser/ cosmetic therapy for angiokeratomas/ compression of oedema
Hospital-based and community services and supports	Psychology	Depression and anxiety; Support for chronic disease
Hospital-based and community services and supports	Social Work	Psychosocial support: Assess social and family supports, safeguarding Link with community supports as required e.g GP, Public Health Nurse, Primary Care SW, Local authority SW, Mental Health SW, Disability SW, TUSLA Offer 1-1 counselling or GP referral to Counselling in primary care (CIPC) www.hse.ie/eng/services/list/4/mental-health-services/counsellingpc/

		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/ Housing and/or mobility issues: Advocacy and support - request adapted room if required. Facilitate housing transfer Home Care Packages: if issues with activities of daily living - arrange application for inpatients, liaise with Public Health Nurse to arrange for out-patients Respite Care: liaise with public health nurse or community disability services to arrange Employment issues: Link to Intreo public employment and EmployAbility services www.gov.ie/en/campaigns/fb84c0-intreo/ www.gov.ie/en/service/8578c4-access-the-employability-service/
Hospital-based and community services and supports	Physiotherapy	Heart Failure: Cardiac Rehab Valvular Disease (post-op): Cardiac Rehab Lymphoedema: Lymphoedema management; MLD, multilayer compression bandaging, compression garments TIA/Stroke: Neuro rehabilitation Acroparesthesia: Provision of brace/mobility aid, gait training Osteopenia/Osteoporosis: Exercise Therapy - to enhance bone mineral density (BMD) or slow/prevent loss of BMD Vertigo: Vestibular Rehabilitation – balance and gait training Monitor for Acroparesthesia - can be exacerbated by exercise, stress and fever (rise in body temperature)
Hospital-based and community services and supports	Occupational Therapy	Assessment and intervention for difficulties participating in activities of daily living; including sleep, self-care, productivity and leisure. Environmental assessments and adaptations if required. Postural management assessment and intervention, including specialist seating and equipment provision if required.
Hospital-based and community services and supports	Speech & Language Therapy	Assess and support speech, language, and communication needs (SLCN) and / or feeding, eating, drinking & swallowing (FEDS) skills based on individual needs / priorities TIAs, Stroke, Cognitive decline and dementia (rare): may impact SLCN and FEDS skills Cochlear implants: formal assessment for suitability, assist audiology, auditory training, language, speech and voice rehabilitation (i.e. training), communication skills, liaison with community SLT and post-operative progress monitoring (should occur along normal developmental lines in the absence of additional needs) Resource: www.beaumont.ie/media/Cochlear_Kids_Book_20121.pdf

Primary and community services and supports	GP	Management of inter-current conditions; co-ordination of local services and supports; refer/communication across services
Primary and community services and supports	Home Infusion Team	GP, Pharmacy, Homecare Nurse Specialist
Primary and community services and supports	Patient Organisations	Advocacy, support and information: Fabry Ireland (www.fabryireland.ie), Rare Diseases Ireland (www.rdi.ie), Rare Ireland Family Support Network (www.rareireland.ie) Fabry International Network (www.fabrynetwork.org)

Clinical Lead:

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