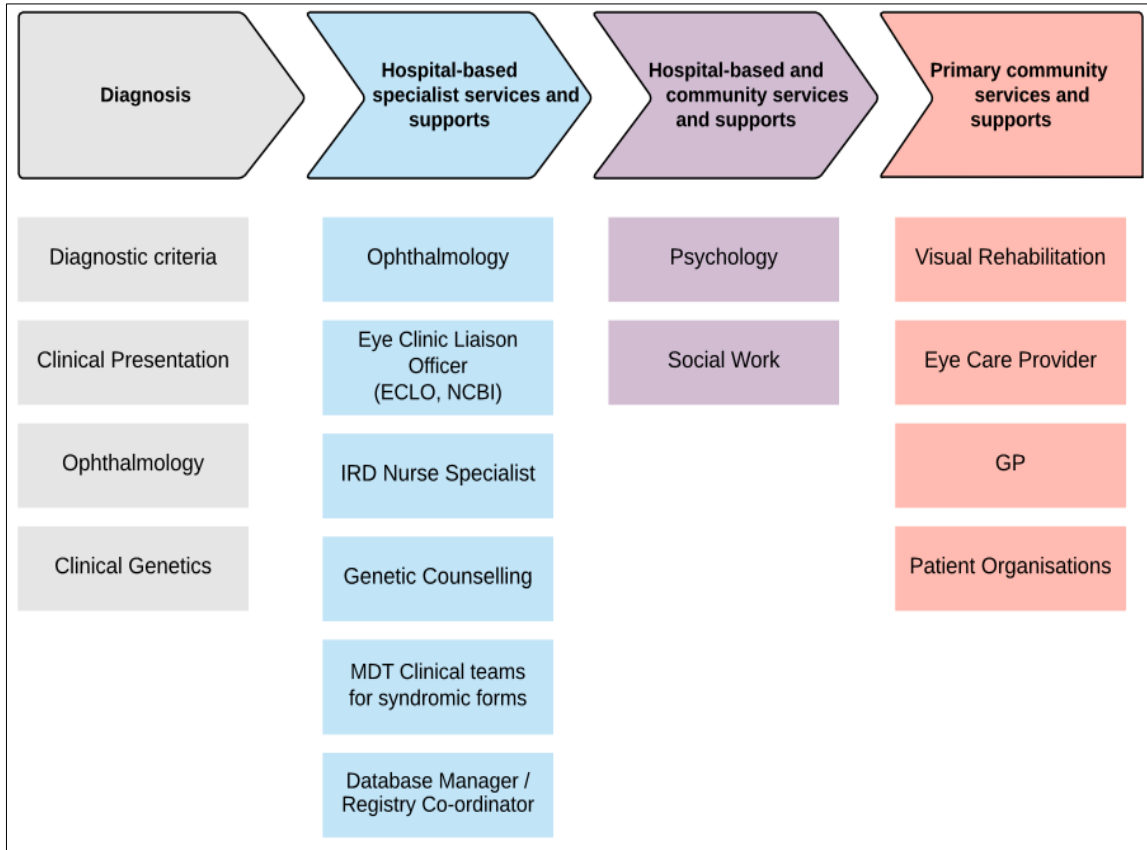




# **Retinitis Pigmentosa (RP)**

## **Adult Care Pathway**

## Retinitis Pigmentosa (RP) Adult Care Pathway



### Clinical Characteristics:

**Orphacode: 791**

**Disease Definition:** Retinitis Pigmentosa (RP) is an inherited retinal dystrophy leading to progressive loss of the photoreceptors and retinal pigment epithelium and resulting in blindness usually after several decades

Diagnosis	Diagnostic Criteria	Consensus (Marmor 1983, Sergouniotis 2019) Rod dysfunction: Dark adaptation (elevated rod final threshold)/ electroretinogram (ERG); progressive loss in photoreceptor function; loss of peripheral vision; bilateral (highly symmetrical). Consider Syndromic vs. Non-syndromic Retinitis Pigmentosa (RP)
Diagnosis	Clinical Presentation	Loss of visual acuity; reduced fields of vision, tunnel vision; blind spots (scotomas); night blindness; photophobia, dysphotopsia
Diagnosis	Ophthalmology	Best corrected visual acuity (BCVA), Snellen charts, Best Corrected Near Acuity (BCNA), +/- Contrast Sensitivity, slit lamp biomicroscopy, intraocular pressure, indirect ophthalmoscopy; Imaging: Colour/wide-field fundus photography, Fundus autofluorescence, optical coherence tomography (OCT); static and kinetic perimetry (visual field); Microperimetry, electroretinogram (ERG) Referral from community eye care provider
Diagnosis	Clinical Genetics	Establishment of Genotype-phenotype correlation. Full systemic examination may be required if syndromic form considered. Gene panels (>280 genes) Irish data (Whelan 2020, Stephenson 2021) - autosomal dominant, autosomal recessive or X-linked, rarely digenic. Diagnostic correlation via Multi-Disciplinary Team (MDT) Meetings
Hospital-based specialist services and supports	Ophthalmology	Eye review 2 yearly with more frequent review if indicated or ocular co-existing conditions (e.g. Cataracts, cystoid macular oedema, glaucoma, epiretinal membrane). Repeat imaging at 2 yearly visit: Color/wide-field fundus photography, Fundus auto-fluorescence, optical coherence tomography (OCT); static (and / or kinetic) perimetry (visual field); Close communication with and provision of education materials for community eye care provider Primary Disease modification with Approved Therapy when available (e.g., for Biallelic RPE65 Disease) Research trials such as gene therapy Offer Eye Clinic Liaison Officer (ECLO) assessment or review
Hospital-based specialist services and supports	Eye Clinic Liaison Officer (ECLO, NCBI)	Initial point of contact for newly diagnosed patients and their families in clinic

		<p>Provide emotional and practical support to help patients and families deal with the diagnosis and maintain independence</p> <p>Work closely with the voluntary support agencies in the community</p> <p>Advise and signpost on the broad range of services available, such as visual rehabilitation, assistive technology, employment support, orientation and mobility, well-being and emotional support and access to other necessary services</p>
Hospital-based specialist services and supports	IRD Nurse Specialist	<p>Patient contact and advice</p> <p>Coordination of services and supports</p> <p>Patient and family education and support</p> <p>Transition planning</p> <p>Liaison with other health care professionals</p> <p>Link families to patient organisations</p>
Hospital-based specialist services and supports	Genetic Counselling	<p>Counsel on mode of inheritance and recurrence risk</p> <p>Autosomal Dominant &amp; X-linked - test other affected family members, identify at-risk relatives, offer genetic counselling and predictive testing for the familial pathogenic variant, as appropriate</p> <p>Autosomal Recessive - recurrence risk estimate</p> <p>Genetic counselling offered within 3-6 months of diagnosis</p>
Hospital-based specialist services and supports	MDT Clinical teams for syndromic forms	<p>Access to national and ERN MDT networks</p>
Hospital-based specialist services and supports	Database Manager / Registry Co-ordinator	<p>Create and maintain database of patients attending service</p> <p>Record patient biographical, clinical and research data</p> <p>Audit and quality improvement</p> <p>Ensure minimum data set standards for European Reference Network (ERN) registry</p>
Hospital based and community services and supports	Psychology	<p>Support for chronic disorder and visual loss</p> <p>Psychological counselling via National Council for the Blind Ireland</p>
Hospital based and community services and supports	Social Work	<p>Access to hospital based social worker referral from Ophthalmology team</p>

Psychosocial support: Assess social and family supports, safeguarding

Link with community supports as required e.g., GP, Public Health Nurse, Primary Care Social Worker, Local authority Social Worker, Mental Health Social Worker, Disability Social Worker, 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/](http://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/](http://www.citizensinformation.ie/)

Housing and/or mobility issues: Advocacy and support

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:

Intreo public employment

[www.gov.ie/en/campaigns/fb84c0-intreo/](http://www.gov.ie/en/campaigns/fb84c0-intreo/)

EmployAbility services

[www.gov.ie/en/service/8578c4-access-the-employability-service/](http://www.gov.ie/en/service/8578c4-access-the-employability-service/)

Primary and community services and supports

Visual Rehabilitation

Access via NCBI, referral from Ophthalmology and ECLO Assessment and intervention action plan and training for difficulties participating in activities of daily living

Primary and community services and supports

Eye Care Provider

Correct refractive error, lenses with special filters. Close liaison with Ophthalmology wrt diagnosis and care plan

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	Patient Organisations	Advocacy, support, information: National Council for the Blind Ireland ( <a href="http://www.ncbi.ie">www.ncbi.ie</a> ) Fighting Blindness ( <a href="http://www.fightingblindness.ie/">www.fightingblindness.ie/</a> ) Rare Diseases Ireland ( <a href="http://www.rdi.ie/">www.rdi.ie/</a> ) Guide Dogs for the Blind ( <a href="http://www.guidedogs.ie">www.guidedogs.ie</a> ) Rare Ireland Family Support Network ( <a href="http://www.rareireland.ie">www.rareireland.ie</a> ) ChildVision ( <a href="http://www.childvision.ie">www.childvision.ie</a> )

### Clinical Leads:

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