

Rare Genetic Disorders and the Medical Health Care Professional

PATH40010

Undergraduate Medicine Programme Detail

YEAR

Wk	Semester 1															Holiday		Semester 2															Summer					Credits																					
	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	16	Christmas	Christmas	17	18	19	20	21	22	23	24	25	26	27	28	29	30	31	32	33	34	35		36	37	38	39	40	41	42	43	44	45	46	47	48	49	50	51					
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Rare Genetic Disorders and the Medical Healthcare Professional (PATH40010)

- In Program Elective module
- Stage 3, semester 2 (undergraduate program)
- January 2012 -
- Class size limited to 35









Module descriptor

- “There is a lack of awareness among medical professionals about rare conditions and more often than not, the diagnosis of these disorders in individuals is a long and gruelling process for the patient”
- “Ireland is required to have a rare disease strategy in place by 2013 under an EU Council recommendation and all government parties have pledged to introduce a national strategy for rare disorders.”
- “In line with this, this module is designed to enhance awareness of rare genetic disorders among future medical professionals.”

Learning Outcomes

1. Understand what is meant by the term “**rare disease**”
2. For selected rare genetic disorders, demonstrate scientific knowledge on the **inheritance patterns, mechanisms and treatment options**
3. Be aware of the **ethical considerations** associated with managing patients and their families with rare genetic disorders
4. Reflect on the **obstacles and difficulties** encountered by patients with rare disorders
5. Appreciate the **importance of research** to diagnosis and treatment of rare diseases

6. Recognise the importance of identifying **novel treatments** for these orphan genetic disorders
7. **Be informed** about current Governmental Health Policies and funding initiatives to support Rare Disease Research.
8. Be able to identify the **information resources** available to them
9. **Recognise the possibility that they may come across a rare disease at some stage in their careers**

 General Info
  Description, Outcomes
  **Workload FTE**
 Prior Learning
  Requirements / Exclusions
  Assessment
  Resits and Remediation
  Assoc Majors
  Derogations


Workload and FTE

Workload

Code	Workload
Autonomous Student Learning	80
Lectures	24
Total Workload	
	104

FTE Equivalents

School	% Ownership
Medicine & Medical Science	100

 **Back**

Assessment

- Continuous Assessment
 - Reflective journal – 50%
 - Prepare a document for medical healthcare professional about rare disorders detailing clinical example – 50%
- Pass/Fail module

Rare Disorders and the Medical Healthcare Professional (PATH40010)

Lecture Timetable 2011 / 2012

		Lecture	Date	Time	Lecturer
wk1	L1	Introductory Lecture	Tues 17 Jan	3pm	Dr Paula Byrne UCD
	L2	Rare Disorders in Ireland	Thurs 19 Jan	4pm	IPPOSI
wk 2	L3	Living with Epidermolysis Bullosa	Tues 24 Jan	3pm	Patient Educator, EB nurse, DEBRA Ireland
	L4	Mechansims and Therapeutics in EB	Thurs26 Jan	4pm	DEBRA Ireland
wk 3	L5	Drug Development Strategies for Rare Disorders	Tues 31 Jan	3pm	Professor of Medicine
	L6	Clinical Trials	Thurs 2 Feb	4pm	to be confirmed
wk 4	L7	α -1 Antitrypsin Deficiency - Clincial presentation, Diagnosis and Treatment	Tues 7 Feb	3pm	Alpha-1 Foundation
	L8	Self Directed Study	Thurs 9 Feb	4pm	
wk 5	L9	Rare Disorders in General Practice	Tues 14 Feb	3pm	to be confirmed
	L10	Genetic Counselling for Rare Disorders	Thurs16 Feb	4pm	Genetic Counsellor
wk 6	L11	Fabry Disease - "Diagnosis is a Journey"	Tues 21 Feb	3pm	Patient Educator
	L12	Enzyme Replacement Therapy	Thurs23 Feb	4pm	Pharmaceutical Industry
wk 7	L13	Living with 22q11 Deletion	Tues 28 Feb	3pm	Patient Educator/Consultant Physician
	L14		Thurs 2 Mar	4pm	
		Study Break			

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Lecture Timetable 2011 / 2012

		Lecture	Date	Time	Lecturer
		Study Break			
wk 8	L14	Rare Disorders in the Irish Travelling Community	Tues 20 Mar	3pm	Medical Geneticist
	L15	Approaches to Mapping Rare Disease Genes	Thurs 21 Mar	4pm	Research Scientist
wk 9	L16	Living with Retinitis Pigmentosa	Tues 27th Mar	3pm	Fighting Blindness
	L17	Mechanisms and Therapeutics for RP	Thurs 29th Mar	4pm	Professor of Genetics
wk 10	L18	Sarcoidosis -An Idiopathic Dilemma	Tues 3rd Apr	3pm	Consultant Physician
	L19	Intellectual disability and rare disorders	Thurs 5th Apr	4pm	to be confirmed
wk 11	L20	Access to services and resources	Tues 10th Apr	3pm	to be confirmed
	L22	National and EU policy for rare disorders/ General discussion and feedback session	Thurs 12th Apr	4pm	to be confirmed
wk 12	L23	Self directed study	Tues 17th Apr	3pm	
	L24	Self directed study	Thurs 19th Apr	4pm	