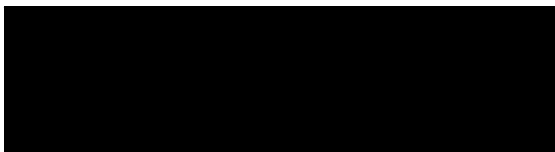


CURRICULUM VITAE

JOANNE HUGHES

Joanne Hughes

Personal Details



Registration Details

Irish Medical Council:



Specialist Register (paediatrics)

General Medical Council (UK):



Full registration with a licence to practice
On specialist register for Paediatrics

Academic Qualifications

Feb 2000	MRCPCH	London
July 1996	MRCP (Paed) Part 1	London
July 1992	MB, BCh, BAO	Queen's University, Belfast

Scholarships

SSIEM Symposium Travelling Scholarship 2005
Irish American Paediatric Society Travel Scholarship 2005

Membership of Professional Societies

Royal College of Paediatrics and Child Health (RCPCH)
Society for the Study of Inborn Errors of Metabolism (SSIEM)
British Inherited Metabolic Diseases Group (BIMDG)
Society for Inherited Metabolic Disorders (SIMD)



Current Employment

01st July 2011 - present

Consultant Paediatrician with an interest in Inherited Metabolic Disorders

National Centre for Inherited Metabolic Disorders
Children's University Hospital, Temple Street, Dublin 1

I am the one of a team of four metabolic consultants in the NCIMD. I work 3 days in the NCIMD and 2 days in Our Lady's Children's Hospital, Crumlin.

Duties include:

- Weekly metabolic clinic in NCIMD
- Quarterly Metabolic Outreach Clinic in Limerick
- Weekly neuroradiology meeting
- Weekly Grand rounds
- Weekly meetings with laboratory staff
- Metabolic consult service to OLCHC with a view to establishing a more robust service.
- Metabolic on-call 1:4
- Medical representative on Clinical Governance Committee

25th Feb 08 – 30th June 2011

Consultant Paediatrician with an interest in Inherited Metabolic Disorders

Royal Belfast Hospital for Sick Children

This was a new post providing a regional service for patients with Inherited Metabolic Diseases.

Duties include:

- Lead clinician paediatric metabolic medicine.
- Weekly metabolic clinic in conjunction with a Clinical Chemist
- Weekly neuroradiology meeting,
- Monthly neuropathology meeting
- Fortnightly meetings with laboratory staff to review clinical cases and lab results
- Twice yearly Mucopolysaccharidoses clinic
- Quarterly Fabry clinic
- Reviewing protocols and emergency regimen for metabolic patients
- Liaising with Newborn screening lab and first contact for positive results
- General paediatric on-call 1:6
- General paediatric review clinic
- Teaching junior medical staff in RBHSC and other paediatric hospitals in Northern Ireland on Metabolic Disorders
- Chair of RBHSC Junior Doctors Committee
- Clinical supervisor to junior medical staff

Fellowship

22nd Jan 07 to 31st Dec 07

12 Months

1:2 metabolic on-call

Prof John Christodoulou

Fellow in Genetic Metabolic Medicine

Children's Hospital at Westmead, Corner of Hawkesbury Rd and Hainsworth St, Locked bag 4001, Westmead, NSW 2145

- First on-call to a busy tertiary referral unit for acute management and elective investigation of patients with inborn errors of metabolism
- Consultations to inpatients at the request of treating paediatricians
- Outpatient clinics include:
 - PKU
 - General Metabolic
 - Mucopolysaccharide
 - RETT Management
- Follow-up for all patients screened positive in the Newborn screening lab
- ERT for children and adults with lysosomal disorders
- Participation in weekly academic meetings
 - Clinical genetics journal club
 - Biochemical genetics journal club
 - Perinatal meeting
 - Neurogenetic radiology meeting
 - Case preparation for Fellows teaching
- Honorary lecturer at the University of Sydney

SpR Appointments

1st Jan 05 to 31st Dec 06

24 Months

1:4 Metabolic on-call

1:10 General paed on-call

SpR Paediatric Metabolic Medicine

National Centre for Inherited Metabolic Diseases, Children's University Hospital, Temple Street, Dublin

4th Aug 04 to 31st Dec 04

5 months

1:5 on-call NICU, RMH

SpR Community Paediatrics

Cupar Street Clinic, N&W Belfast Trust, 91 Cupar Street, Belfast, BT13 2LJ

4th Feb 04 to 3rd Aug 04

6 Months

1:7 General Paeds on-call

1:10 PICU on-call

SpR Neurology

Royal Belfast Hospital for Sick Children, Falls Road, Belfast BT12 6BA

6th Aug 03 to 3rd Feb 04

6 Months

1:7 general paed on-call

1:10 PICU on-call

SpR General Paediatrics, Endocrinology and Diabetes

Royal Belfast Hospital for Sick Children, Falls Road, Belfast, BT12 6BA

5th Feb 03 to 4th Aug 03

6 months

1:4 on-call

SpR Neonatology

NICU, Royal Maternity Hospital, Grosvenor Road, Belfast, BT12 6BA

28th Oct 02 to 4th Feb 03

3 Months

1:5 on-call NICU, RMH

SpR Community Paediatrics

Cupar Street Clinic, N&W Belfast Trust, 91 Cupar Street, Belfast, BT13 2LJ

Metabolic Clinic, Belfast

30th May 01 to 27th Oct 02
17 Months
1 day/week

Medical officer (Part-time), Metabolic Clinic
 Royal Belfast Hospital for Sick Children, Falls Road, Belfast,
 BT12 6BA

Staff Grade Community Paediatrics

1st Oct 01 to 27th Oct 02
13 Months

Staff Grade Community Paediatrics
 Homefirst Community Trust, 60 Steeple Road, Antrim,
 BT41 2 RJ

1st Nov 00 to 30th Sept 01
11 Months

Staff Grade Community Paediatrics
 Armagh Community Clinic, Tower Hill, Armagh
 BT61 9DR

Middle Grade SHO Appointments

2nd Aug 00 to 31st Oct 00
3 Months
1:5 on-call (Reg roster)

General Paediatrics and Neonatology
 Craigavon Area Hospital, 68 Lurgan Road, Portadown,
 BT63 5QQ

1st Mar 00 to 1st Aug 00
5 Months
1:4 on-call (Reg roster)

SHO (in lieu of REG) Neonatology
 NICU, RMH, Royal Maternity Hospital, Grosvenor Road,
 Belfast, BT12 6BA

2nd Feb 00 to 29th Feb 00
1 Month
1:5 on-call

SHO General Paediatrics
 Daisy Hill Hospital, 5 Hospital Road, Newry, BT35 8DR

4th Aug 99 to 25th Sept 99
2 Months

SHO Clinical Genetics
 Belfast City Hospital, 51 Lisburn Road, Belfast, BT9 7AB

3rd Feb 99 to 3rd Aug 99
6 Months
1:5 on-call (Reg roster)

General Paediatrics and Neonatology
 Ulster Hospital Dundonald, Upper Newtownards Road,
 Dundonald, BT16 1RH

Paediatric SHO Appointments

31st Aug 98 to 2nd Feb 99
5 Months
1:5 on-call

SHO Neonatology
 Jubilee Maternity Hospital, Belfast City Hospital, 51 Lisburn
 Road, Belfast, BT9 7AB

4th Feb 98 to 25th Apr 98
3 Months
1:5 on-call Neonatology

SHO Clinical Genetics
 Belfast City Hospital, 51 Lisburn Road, Belfast, BT9 7AB

6th Aug 97 to 3rd Feb 98
6 Months
1:5 on-call

SHO General Paediatrics
 Daisy Hill Hospital, 5 Hospital Road, Newry, BT35 8DR

5th Feb 97 to 5th Aug 97
6 Months
1:4 on-call

SHO Neonatology
 NICU, Royal Maternity Hospital, Grosvenor Road, Belfast,
 BT12 6BA

4th Dec 96 to 4th Feb 97
2 Months
Partial shift rota

SHO Paediatric Surgery

Royal Belfast Hospital for Sick Children, Falls Road, Belfast,
BT12 6BA

2nd Oct 96 to 3rd Dec 96
2 Months
Partial shift rota

SHO General Paediatrics

Royal Belfast Hospital for Sick Children, Falls Road, Belfast,
BT12 6BA

7th Aug 96 to 1st Oct 96
2 Months
Full shift rota

SHO Accident and Emergency Dept

Royal Belfast Hospital for Sick Children, Falls Road, Belfast,
BT12 6BA

2nd Aug 95 to 6th Aug 96
12 Months
1:4.5 on-call

SHO General Paediatrics and Neonatology

Antrim Area Hospital, 45 Bush Road, Antrim, BT41 2RL

Medical SHO Appointments

1st Feb 95 to 1st Aug 95
6 Months

SHO Neurology

Royal Victoria Hospital, 274 Grosvenor Road, Belfast,
BT12 6BA

1st Aug 94 to 31st Jan 95
6 Months

SHO Endocrine, Gastroenterology and General Medicine

Belfast City Hospital, 51 Lisburn Road, Belfast, BT9 7AB

1st Feb 94 to 31st July 94
6 Months

SHO Rheumatology

Musgrave Park Hospital, Stockman's Lane, Belfast, BT9 7JB

1st Aug 93 to 31st Jan 94
6 Months

SHO General Medicine, Cardiology, Paediatrics and Geriatrics

Tyrone County Hospital, Hospital Road, Omagh,
BT79 0AP

Pre-registration

1st Apr 93 – 31st July 93
4 Months

JHO Nephrology

Belfast City Hospital, 51 Lisburn Road, Belfast, BT9 7AB

1st Dec 92 – 31st Mar 93
4 Months

JHO General Medicine

Downe Hospital, 9A Pound Lane, Downpatrick, BT30 6JA

1st Oct 92 – 30th Nov 92
2 Months

JHO Urology

Belfast City Hospital, 51 Lisburn Road, Belfast, BT9 7AB

1st Aug 92 – 30th Sept 92
2 Months

JHO Vascular and General Surgery

Belfast City Hospital, 51 Lisburn Road, Belfast BT9 7AB

Publications

Joanne Hughes, Stephanie Ryan, Deborah Lambert, Olivia Geoghegan, Anne Clark, Yvonne Rogers, Una Hendroff, Ahmad Monavari, Eilish Twomey and Eileen Treacy **“Outcomes of Siblings with Classical Galactosemia”** *The Journal of Pediatrics*, Volume 154, Issue 5, May 2009, Pages 721-726

JAF Hughes, C Stenson, M O Regan, M Macauley, M Irranca, A Monavari, EP Treacy. **“Maternal Phenylketonuria: An Irish perspective”** *Journal of Inherited Metabolic Disease* 2006, 29: (Supplement 1), 97

HB Chew, O Walsh, JAF Hughes, S Mohammed, M Irranca, A Clark, C O Neill, A Monavari, EP Treacy, R Manning. **“3 Successful Spontaneous Pregnancies in 2 Women with Classical Galactosaemia”** *Journal of Inherited Metabolic Disease* 2006, 29: (Supplement 1), 124

RM Brown, RA Head, J Hughes, MU Halldin, AE Slonim, SH Green, A Chakrapani, GK Brown. **“Clinical and genetic heterogeneity of dihydrolipoamide dehydrogenase deficiency”** *Journal of Inherited Metabolic Disease* 2006, 29: (Supplement 1), 1

J O'Connor, C McDonnell, JAF Hughes, R Manning **“A Study on Growth in Children with Classical Galactosaemia”** *Irish Journal of Medical Science* Nov 2006; 175 (4) es 2:34

J Hughes, A Clark, O Geoghegan, J O Toole, U Hendroff, Y Rogers, O Walsh, M O Regan, C Steenson, D Lambert, S Yap, R Manning, E Treacy. **“A study on outcomes in siblings with classical Galactosaemia in Ireland”** *Journal of Inherited Metabolic Disease* 2005, 28: (Supplement 1), 206.

J Hughes, E Treacy. **“Maple Syrup Urine Disease”** *Medicine Weekly* (14th Sept 2005)

PJ Morrison, D McManus, J Hughes, NC Nevin, D Webb. **“Subcortical white-matter abnormalities due to a previously undescribed de novo 14q duplication defined by FISH”** *Developmental Medicine and Child Neurology* 1998, 40 :(Supplement 79), 25