SMA Treatment Network

Education Event

28th November 2022 13:30 – 16:45

Introduction

SMA is a treatable condition. Treatment is more effective with earlier diagnosis, so SMA is now a time critical diagnosis.

Part A - The importance of early diagnosis

Primary audience: All involved in SMA care, with a focus on health visitors, general and community paediatricians, GP trainees, neonatologists.

1:35	**Case studies of new presentations** *Here we will explore 2 different cases of children who ultimately received a diagnosis of SMA, how the diagnostic journey evolved, the experience of the healthcare professionals, and the outcomes for each child*	**Presenters:** Dr Diarmuid Kerrin, Barnsley Hospital NHS Foundation Trust Dr Gary McCullagh, Royal Manchester Children's Hospital
1:55	**What to look for** *Here we take a look at what best practice tells us about how to identify suspect SMA, with the aim of arming professionals involved in the care of young children with the right skills to make timely referral.* *We'll also share the experience of a particular family and their journey.*	**Presenter:** Rajesh Madhu, Alder Hey Children's Hospital
2:20-2:50	**New born screening** *Here we hear discuss the effectiveness of new-born screening on pre-symptomatic patients, pilots underway and the approaches different countries have taken.*	**Presenter:** Dr Sithara Ramdas, Oxford University Hospitals NHS Foundation Trust
2.50-3.00	**Questions & discussion**	
3.00-3.10	**Comfort break**	

RCPCH has approved this activity for CPD in accordance with the current RCPCH CPD guidelines.

This meeting has been supported by Novartis Gene Therapies via provision of funding to cover use of a virtual event platform and logistical support. Novartis Gene Therapies had no involvement in the agenda or speaker selection.

Part B - Treatment and management

Primary audience: All involved in SMA care, likely of special interest to those involved in the later elements of the pathway such as consultant neurologists/paediatricians, specialist nurses, Allied Health Professionals.

3:10-3.35	**Respiratory care** *Here we will explore the importance of respiratory assessment and management, develop comprehensive awareness of respiratory management including prevention, secretion clearance, ventilatory support, management of acute exacerbations*	**Presenter:** Dr Stuart Wilkinson, Royal Manchester Children's Hospital
3:35-4.00	**Respiratory physiotherapy and secretion management** *Here we'll explore the topics of secretion management and augmented cough in the SMA patient*	**Presenter:** Sinead Warner and Jasmine Hall, Royal Manchester Children's Hospital
4:00-4.30	**The evolving phenotype of the SMA population** *First we'll have a short talk on the recent development of treatments is extending the lives of children with SMA, and how this changes the phenotypic landscape. We'll then have an open discussion with attendees on the implications for care.*	**Lead:** Anne-Marie Childs, Leeds Teaching Hospitals NHS Trust
4.30-4.45	**Questions & discussion**	

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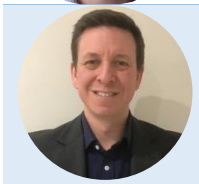
Speaker profiles

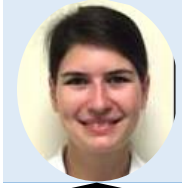
Dr Imelda Hughes, Consultant Paediatric Neurologist
Royal Manchester Children's Hospital - Manchester University NHS Foundation Trust
Having completed paediatric training in Belfast and Bath, and Paediatric Neurology training in Southampton, Dr Hughes has been a Consultant Paediatric Neurologist and Lead Clinician for the Neuromuscular Service at Royal Manchester children's Hospital since 1996.

Dr Gary McCullagh, Consultant Paediatric Neurologist
Royal Manchester Children's Hospital - Manchester University NHS Foundation Trust
Dr McCullagh is a Consultant Paediatric Neurologist with expertise in paediatric neuromuscular disorders. Having initially trained in paediatrics in Belfast, he completed paediatric neurology training in Manchester. Following this, he completed a fellowship in neuromuscular disorders at the Dubowitz Neuromuscular Centre, Great Ormond Street Hospital, London.

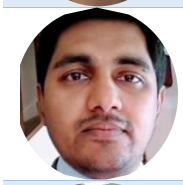


Dr Stuart Wilkinson, Consultant in Paediatric Respiratory Medicine
Royal Manchester Children's Hospital - Manchester University NHS Foundation Trust
Dr Wilkinson graduated from Leicester University in 2003 and trained in paediatrics and respiratory medicine throughout the north in Stoke-on-Trent, Liverpool (Alder Hey) and Sheffield Children's Hospital, before completing sub-specialty respiratory paediatric training in Leicester. He spent 2 years as a cystic fibrosis clinical fellow researching fungal disease in cystic fibrosis before taking a consultant post at Royal Manchester Children's Hospital in 2014.

Sinead Warner, Neuromuscular Physiotherapist
Royal Manchester Children's Hospital - Manchester University NHS Foundation Trust
Sinead Warner completed a physiotherapy degree in Manchester before moving to New Zealand to complete rotational posts including paediatric specialities. On returning to Manchester Sinead worked as a paediatric community physiotherapist developing an interest in neuromuscular conditions. Sinead now works as a highly specialised physiotherapist.

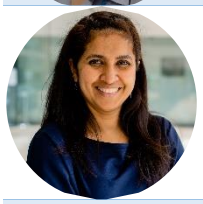


Jasmine Hall, Neuromuscular Physiotherapist
Royal Manchester Children's Hospital - Manchester University NHS Foundation Trust
Jasmine Hall completed a physiotherapy degree at Keele University and completed a range of acute rotations as a junior before specialising in respiratory areas. Jasmine developed an interest in paediatric respiratory conditions and has worked at Royal Manchester Children's Hospital as a Highly Specialised Paediatric Respiratory Physiotherapist since 2018.



Dr Rajesh Madhu, Consultant in the Neurosciences Department
Alder Hey Children's NHS Foundation Trust
Dr Madhu is the Clinical Lead for the Neuromuscular Services at Alder Hey Hospital. He completed his Master's degree in Genomic Medicine from the University of Manchester and leads the Genomic Medicine service for Neuromuscular Disorders. They provide advanced diagnostic investigations for children with suspected neuromuscular disorders including whole genome sequencing.

Dr Diarmuid Kerrin, Consultant Paediatrician
Barnsley Hospital NHS Foundation Trust
Diarmuid Kerrin is a general paediatrician in Barnsley Hospital, a typical DGH. He has been in this post for over 20 years, has interests in atopic disease, cystic fibrosis and safeguarding, and runs weekly allergy clinics. He aims for a pragmatic and family-centred approach, with communication and shared decision-making being key. However, he has little direct experience of SMA



Sithara Ramdas, Consultant Paediatric Neurologist
Oxford University Hospitals NHS Foundation Trust
Sithara Ramdas is a Consultant Paediatric Neurologist and Honorary Senior Clinical Lecturer with the University of Oxford. She completed her paediatric neurology and neuromuscular training at Evelina London Children's Hospital. She is the lead for paediatric neuromuscular service in Oxford and paediatric lead for the NHSE highly specialist service for congenital myasthenia syndromes and neuromyelitis optica.

Anne-Marie Childs, Consultant Paediatric Neurologist
Leeds Teaching Hospitals NHS Trust
Dr Childs has worked as a consultant paediatric neurologist in Leeds since 2001 after becoming the Lead for Children's Neuromuscular Services. Dr Childs is committed to delivering high-quality care to all children and young people with neuromuscular disorders in Yorkshire thus works closely with colleagues in other disciplines, with charities and the UK NM consortium. She is a member of the SMA working group and ethical committee. Clinical research interests include MR imaging and rare disease phenotyping, with growing research in relation to DMD and SMA.

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