

CRC National Specialist Services (NSS)

# Hereditary Motor & Sensory Neuropathy (HMSN)

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# Learning Outcomes

- Understand what HMSN is
  - Aetiology
  - Types of HMSN
  - Pathophysiology
- Understand what happens in the HMSN clinic and how the CRC team can support you
- Orthotics
- Orthopaedics
- Understand the referral pathway

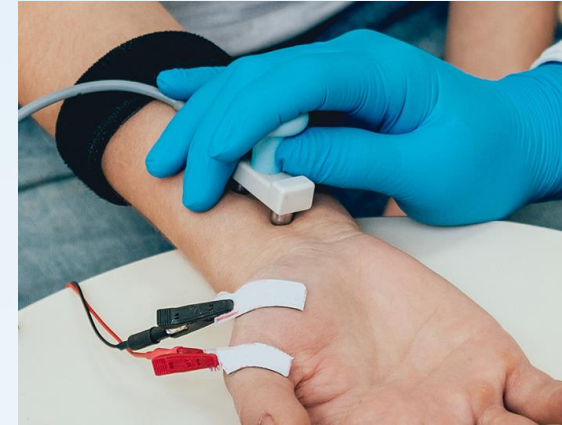
# Aetiology of HMSN

HMSN is also known as Charcot Marie Tooth (CMT)

Primarily caused by gene mutations affecting the peripheral nerves

Inherited condition which is inherited in multiple ways (autosomal dominant, autosomal recessive and x-linked recessive). Multiple Types of HMSN

Diagnosed via symptoms, genetic and nerve conduction tests



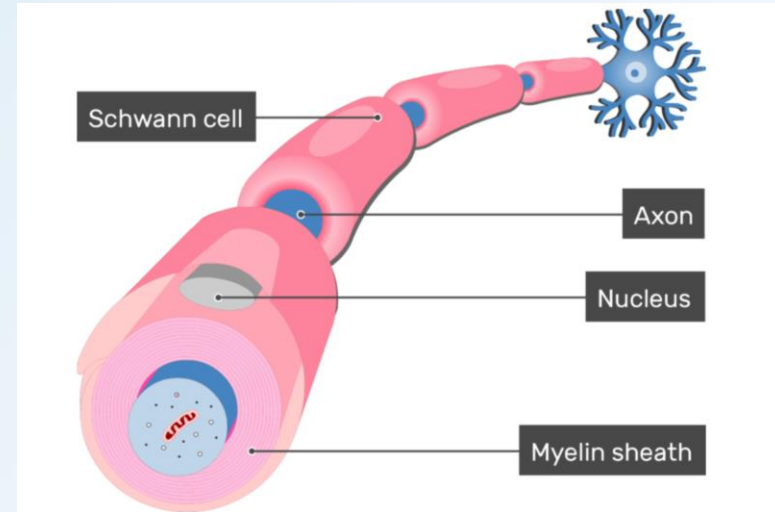
# Pathophysiology of HMSN

- Impact on peripheral nerves
- Type of HMSN can cause either:
  - Demyelination – damaged myelin sheaths
  - Axonal degradation – damaged axons
- Muscle weakness primarily in the feet, ankles and hands
- Sensory loss in the extremities; numbness, tingling, reduced sensation
- Foot deformities; Pes Cavus or Pes Planus
- Abnormal gait (increased trips & falls)
- Decreased balance and stability
- Difficulty with fine motor skills



# Types on HMSN

- Type 1 a/b
  - Demyelinating
  - Most common type
- Type 2
  - Axonal
  - Less severe symptoms than Type 1
- Type 3-7
  - Rare & more severe symptoms
- X-linked
  - Caused by mutation in x chromosome
  - More common in male than female



# HMSN Clinics

A National Consultant led MDT Clinic  
6 Clinics per year, Mon morning, in Clontarf


Consultant Neurologist Dr Declan O'Rourke  
Senior Physiotherapist  
Senior Occupational Therapist  
MDI Liaison Officer

A National Consultant led Orthopaedic/Orthotic Clinic  
3/4 clinics per year, Wed afternoon, in Clontarf

Clinical Specialist Orthotist, Donna Fisher  
Consultant Orthopaedic Surgeon Mr O'Sullivan  
Senior Physiotherapist

# Report

Ref + Client File No.

	<b>HMSN CLINIC</b> <b>Physiotherapy and Occupational Therapy Report</b>
	<b>-Private &amp; Confidential</b> <small>Photocopying or reproduction of this report wholly or in part, requires prior permission of the author.          No responsibility will be accepted for duplicates made without permission.</small>

<b>Name:</b>	<b>Date of Birth:</b>	<b>Current Age:</b>
<b>Address:</b>	<b>Diagnosis:</b>	Years Months
<b>Date of Assessment:</b>	<b>CRC File Number:</b>	
<b>Local Services:</b> <i>Physiotherapist:</i> <i>Occupational therapist:</i>	<b>Local Services - School</b>	

<b>Team members seen today</b>	~ Dr Declan O'Rourke, Consultant Paediatric Neurologist ~ Clodagh Coman, Senior Physiotherapist ~ Laura Lipparoni, Senior Physiotherapist ~ Karen Lennon, Senior Occupational Therapist ~ Donna Fisher, Senior Orthotist ~ Other
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<b>Summary</b>	
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<b>Plan / Recommendations</b>	1. 2.
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<b>Update from family</b>	
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Ref + Client File No.

<b>Activities of Daily life</b>	<i>Self-Care</i> <i>Dressing:</i> <i>Grooming:</i> <i>Bathing/Showering:</i> <i>Feeding:</i> <i>Sleeping:</i>
<b>Productivity</b>	<i>School (+PB?):</i> <i>Hobbies:</i> <i>SNA/Resource:</i> <i>Handwriting/AT:</i>

<b>Symptoms</b>	Foot pain <input type="checkbox"/> Leg cramp <input type="checkbox"/> Unsteady ankle <input type="checkbox"/> Trips and Falls →    Frequently <input type="checkbox"/> Daily <input type="checkbox"/> Hand Pain <input type="checkbox"/> Hand Weakness <input type="checkbox"/> Hand Tremor <input type="checkbox"/>
<b>Comment:</b>	

<b>Sensory Symptoms</b>	<i>(e.g pins&amp; needles; tingling; numbness; prickling)</i>
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Sensations	0	1	2	3	4	Score
<b>Pinprick</b>	Normal	Decreased below or at ankle boots	Decreased at or below midline calf	Decreased at calf midline up to including knee	Decreased above knee (above top of patella)	
<b>Vibration</b>	Normal	Reduced at first metatarsal bone	Reduced at ankle	Reduced at knee (tibial tuberosity)	Absent at knee and ankle	
<b>Temperature</b>	Normal	Reduced at first metatarsal bone	Reduced at ankle	Reduced at knee (tibial tuberosity)	Absent at knee and ankle	

<b>Medical Review</b>	
<b>Foot Posture</b>	<i>Insert picture</i>
<b>Foot condition</b>	
<b>Standing Posture:</b>	
<b>Orthotics</b>	

Ref + Client File No.

Range of Motion	Right*	Left*	Comments
Hip Flex/Thomas Test			
Popliteal Angle			
Ankle Knee Flexed			
Ankle Knee Ext			

Strength	Right	Left	Comments
<b>Hip</b>			
Flexors			
Extensors			
Abductors			
Adductors			
<b>Knee</b>			
Flexors			
Extensors			
<b>Ankle</b>			
Dorsiflexors			
Plantarflexors			

<b>Mobility</b>	<b>Gait Pattern:</b>			
	Able to heel walk	No <input type="checkbox"/>	Yes <input type="checkbox"/>	Some <input type="checkbox"/>
	Able to toe walk	No <input type="checkbox"/>	Yes <input type="checkbox"/>	Some <input type="checkbox"/>
	Tandem Stand	No <input type="checkbox"/>	Yes <input type="checkbox"/>	Some <input type="checkbox"/>
	Tandem Walk (6 steps)	No <input type="checkbox"/>	Yes <input type="checkbox"/>	
	Jump	No <input type="checkbox"/>	Yes <input type="checkbox"/>	Reps ( )
	Hop	No <input type="checkbox"/>	Yes <input type="checkbox"/>	Reps ( )
SLS (eyes open)	L: (sec)	R: (sec)		
SLS (eyes closed)	L: (sec)	R: (sec)		

Ref + Client File No.

<b>Foot Posture index</b>	<b>Talar head palpation</b>	Left	Right
	Curves above and below		
	Let malleolus		
	Inversion/Eversion of the calcaneus		
	Bulge in the region of the talonavicular joint		
	Congruence of the medial longitudinal arch		
	Abd adduction of the forefoot on a rearfoot (too-many-toes)		
	Total (-12 to +12)		
	<b>6MWT</b>		

# How the clinic team can support you

- Work in consultation with the local team to assess clients and track changes to structure and function
- Attend clinics appointments on behalf of the local team
- Feedback clinic findings / reports to the local team
- Provide support to the local team around client needs, orthotics, surgery (pre and post)

# Orthotics & Orthopaedics

- . Review with MDT
- . Current orthotic status
- . Assessment for new orthotics
  - Renew prescription
  - Cast/measure at clinics
  - Local services – advise on prescription
- . Fitting/delivery at OB Ireland clinic in NOH Cappagh

# Referrals

## Who can refer in?

- Paediatricians, GP's, Physiotherapists

[hmsn@crc.ie](mailto:hmsn@crc.ie)

## Referral Criteria:

- Ages up to 18 years
- Dx of HMSN/Peripheral Neuropathy

# Questions?



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physiosecc@crc.ie

Thank you