

# 20 Years of AMEND

AMEND Annual Patient Information Day 2023



A grid of 12 donuts arranged in three rows and four columns. The donuts feature various glazes and toppings: Row 1 includes a chocolate donut with multi-colored sprinkles, a teal donut with silver sprinkles, a pink donut with white stripes, and a chocolate donut with chopped nuts. Row 2 includes a pink donut with multi-colored sprinkles, a chocolate donut with multi-colored sprinkles, a yellow donut with multi-colored sprinkles, and a pink donut with multi-colored sprinkles. Row 3 includes a chocolate donut with chocolate shavings, a teal donut with white stripes, a chocolate donut with chopped nuts, and a yellow donut with white stripes.

National Doughnut Week starts now!

# However, more importantly.....

The month of May is also:

- National Walking Month



- National Mental Health Awareness Week



- International Clinical Trials Month



- National Share a Story Month

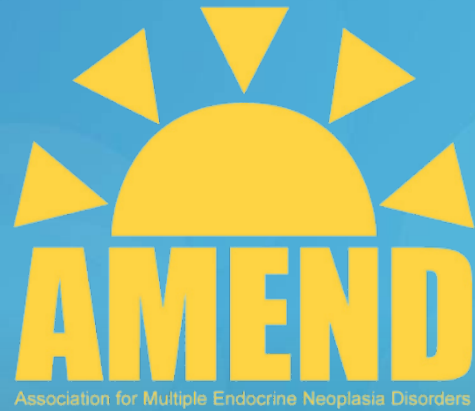


# Agenda

- Housekeeping
- Welcome and introductions
- Overview of AMEND's Patient Communities
- AMEND's 20-years

# Housekeeping

- Fire alarms
- Fire exit
- Toilets
- Lunch and refreshments
- After the conference...

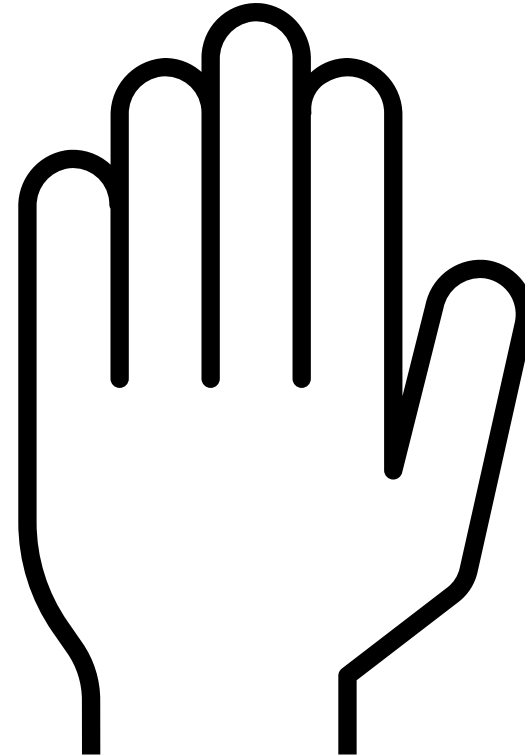


WELCOME!



# Introductions

- Old 'hats'
- Newbies
- Volunteers
- Hangers on?!



## AMEND PATIENT INFORMATION DAY 2023 (#APID2023)

|   |  |  |  |
|---|--|--|--|
| <b>09:30-10:00 REGISTRATION AND REFRESHMENTS</b>  |  |  |  |
| <b>10:00-10:25 INTRODUCTIONS AND HOUSEKEEPING (ROOM 1)</b>  |  |  |  |
| 10:00-10:25 Welcome, Introductions and Housekeeping, Summary Overview of AMEND's 20 Years and our Communities (Jo Grey, AMEND CEO)  |  |  |  |
| <b>10:30-12:30 DISEASE-SPECIFIC SESSIONS</b><br>(Introduction to disease, update on advances in diagnosis of recurrence, imaging, treatments, and research, followed by 1-hour Q&A session) |  |  |  |
| <b>MEN1</b><br><b>(Rooms 3&amp;4)</b><br><i>Chair:</i><br><i>Gill Masters</i><br><b>Medical Expert:</b> <i>Prof Ashley Grossman</i><br><i>(Royal Free London)</i>                           | <b>MEN2A, MEN2B, MTC</b><br><b>(Room 10)</b><br><i>Chair:</i><br><i>Jo Grey</i><br><b>Medical Expert:</b> <i>Prof Karim Meeran</i><br><i>(Imperial London)</i> | <b>PHAEO PARA SYNDROMES</b><br><b>(Room 9)</b><br><i>Chair:</i> <i>tbc</i><br><b>Medical Expert:</b> <i>Dr Ruth Casey</i><br><i>(Addenbrooke's, Cambridge)</i> | <b>Adrenocortical Cancer</b><br><b>(Room 8)</b><br><i>Chairs:</i> <i>Jennifer Rayner</i><br><b>Medical Expert:</b><br><i>Dr Cristina Ronchi</i><br><i>(University of Birmingham)</i> |
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| <b>16:00-17:00 YOGA (Rooms 3+4)</b>   |  |  |  |
| 16:00-17:00 All-ability Beginners' Seated and Unseated Yoga (Lizzie Dunn)   |  |  |  |
| <b>END OF INFORMATION DAY</b>   |  |  |  |





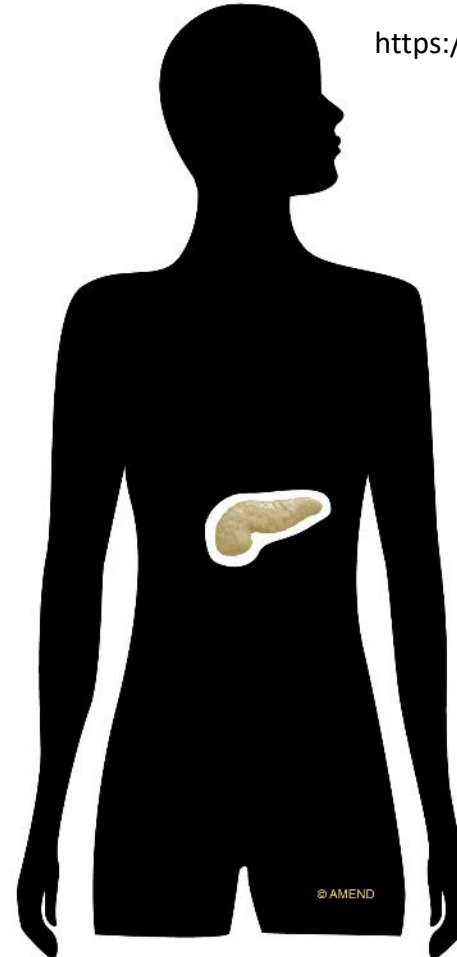
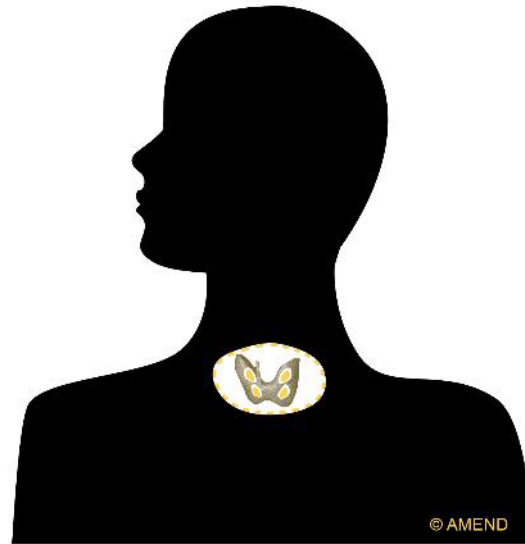
# Overview of AMEND's Patient Communities

- MEN Syndromes
- PPGL Syndromes
- Adrenocortical Cancer
- Medullary Thyroid Cancer

# MEN1 Syndrome (3 Ps)

- Mutation in the MEN1 gene
- Parathyroid adenomas
- Pancreas (panNEN)
- Pituitary (pitNEN)
- Inheritance 50%
- Uncommon

NEN = neuroendocrine neoplasm



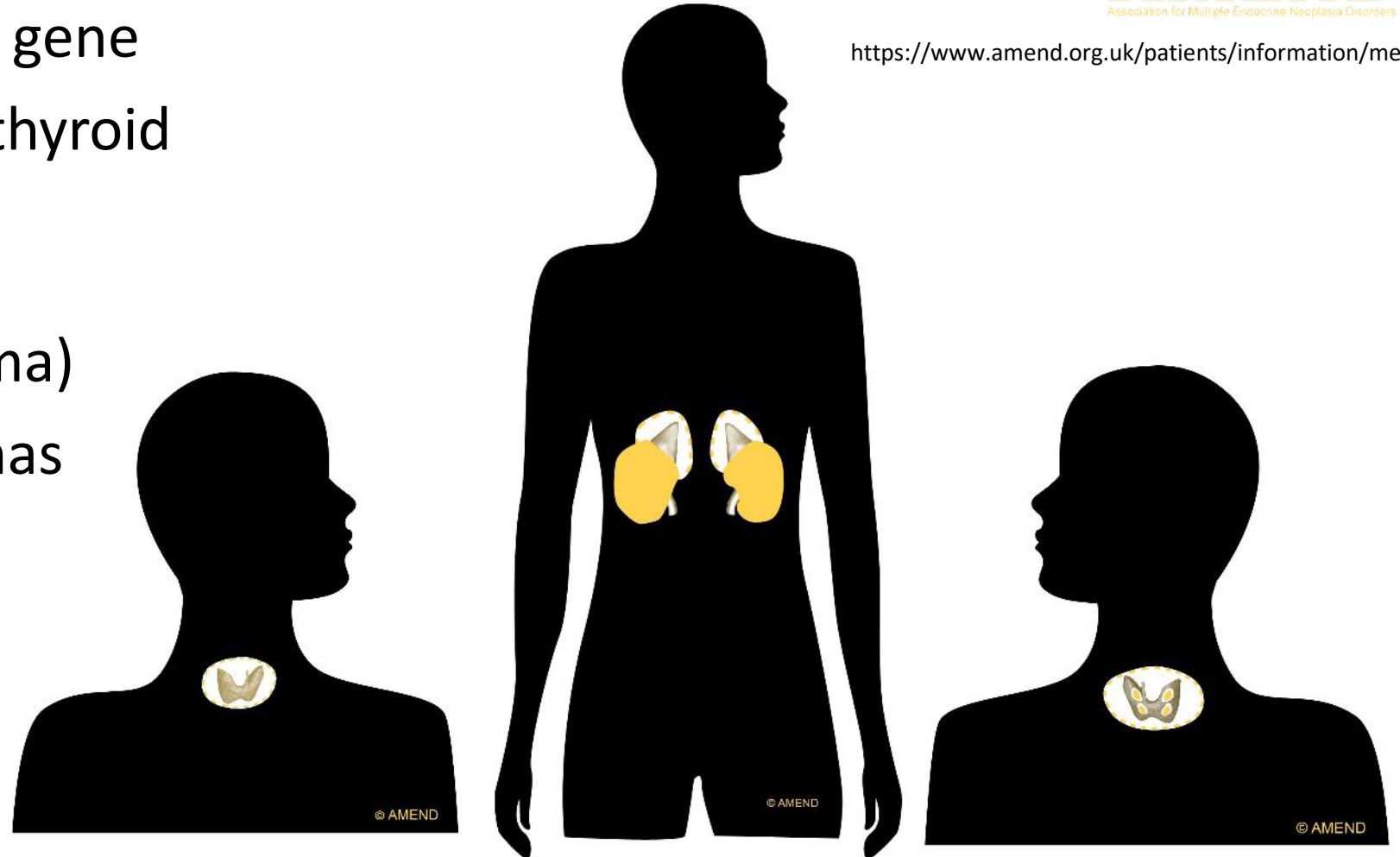
<https://www.amend.org.uk/patients/information/men-1/>

# MEN2 (MEN2A) Syndrome

- Mutation in the RET gene
- Thyroid (medullary thyroid cancer)
- Adrenals (phaeochromocytoma)
- Parathyroid adenomas
- Inheritance 50%
- Rare



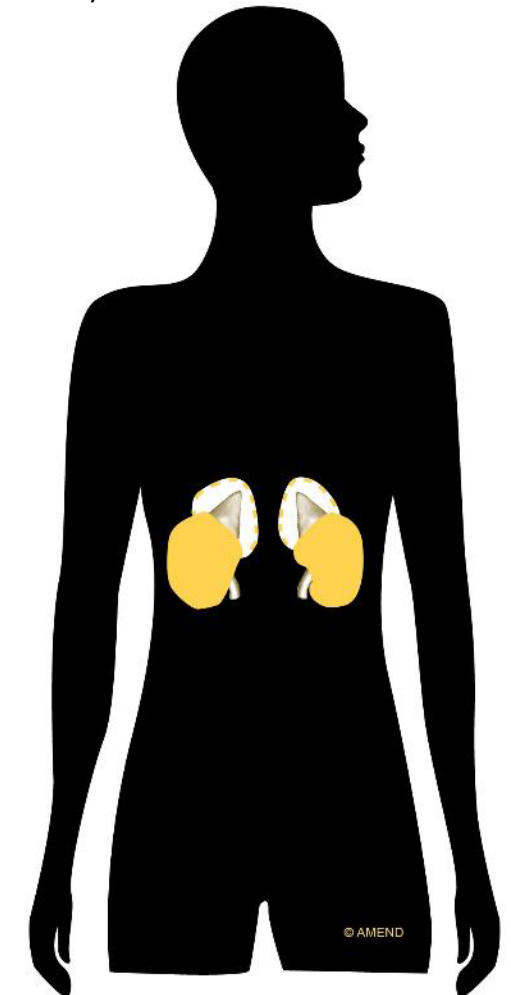
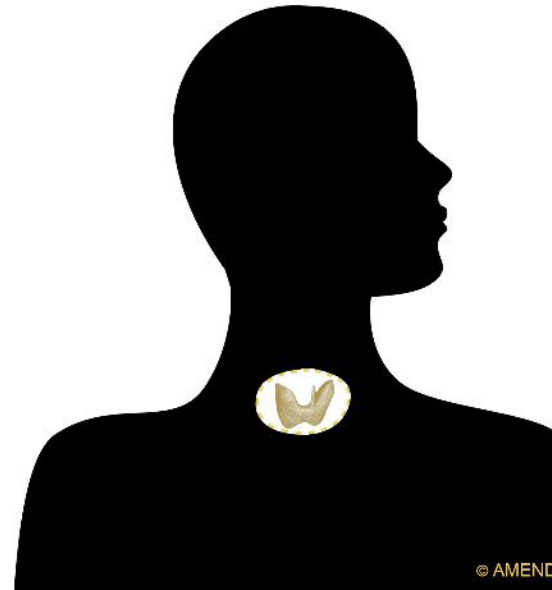
<https://www.amend.org.uk/patients/information/men-2a/>



# MEN3 (MEN2B) Syndrome

- Mutation in the RET gene
- Thyroid (medullary thyroid cancer)
- Adrenals (phaeochromocytoma)
- Gut issues (ganglioneuromatosis)
- Musculoskeletal issues
- Facial differences
- Inheritance 50%
- Most people with MEN2B have new (de novo) mutations
- Extremely rare

<https://www.amend.org.uk/patients/information/men-2b3/>



# PPGL Syndromes

<https://phaeoparasupport.org.uk/>



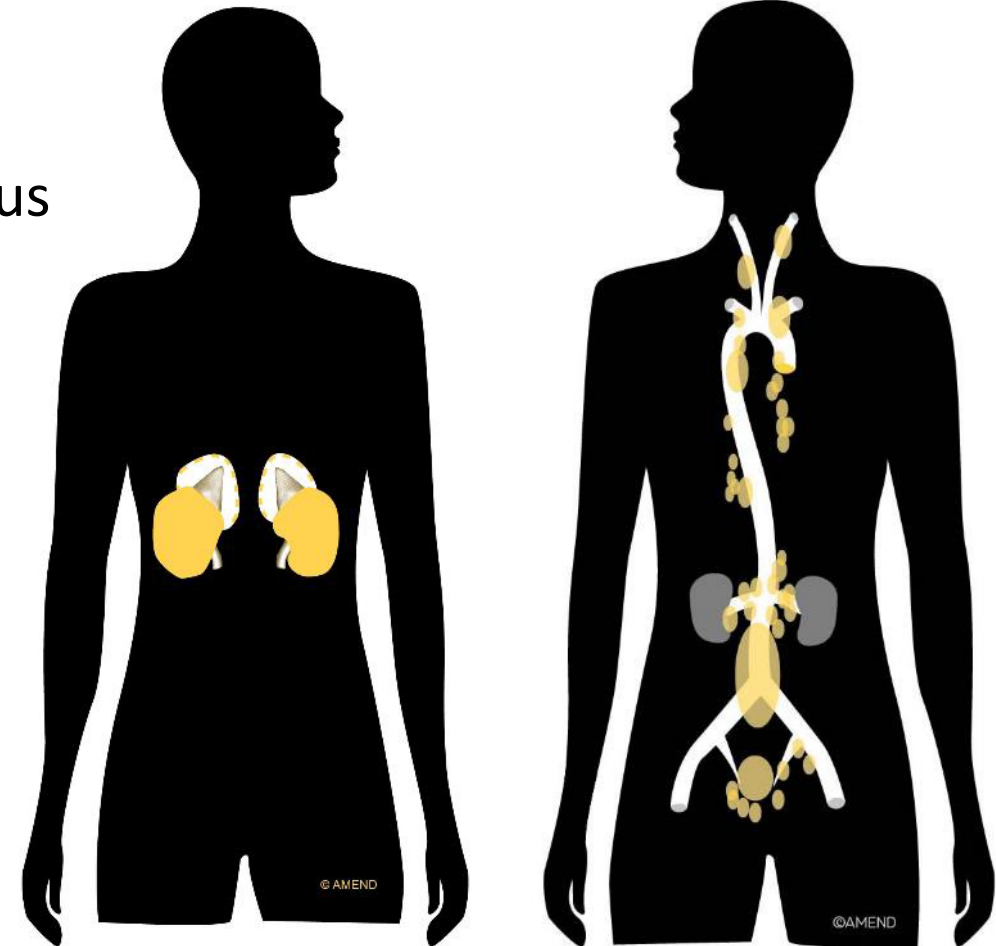
- Adrenals (phaeochromocytoma)
- Paraganglioma
  - Extra-adrenal paraganglia of the nervous system

Table 1: risks of tumours in unaffected individuals with gene mutations

| Gene     | Risk of any tumour (lifetime) | Most likely position of tumour      | Multiple tumours | Other tumours (not PGL/Phaeo) |
|----------|-------------------------------|-------------------------------------|------------------|-------------------------------|
| SDHB     | 25-30%                        | Adrenal and extra adrenal PGL40-50% | 30%              | Renal cell carcinoma<br>GIST  |
| SDHD     | 80-90%**                      | Head and neck                       | 50%              |                               |
| SDHA     | up to 10%                     | Adrenal and extra adrenal PGL       | rarely           | GIST                          |
| SDHAF2*  | 5%                            | Head and neck                       | 90%              | Not reported                  |
| SDHC     | 10%                           |                                     |                  |                               |
| MAX*     | 30-50%                        | Adrenal                             | 60%              | Not reported                  |
| TMEM127* | 40-80%                        | Adrenal                             | 25%              | Not reported                  |

\* Data is limited as numbers of cases are small and therefore the tumour risks are likely to be overestimates

\*\*If the gene change is inherited from the father.



© AMEND

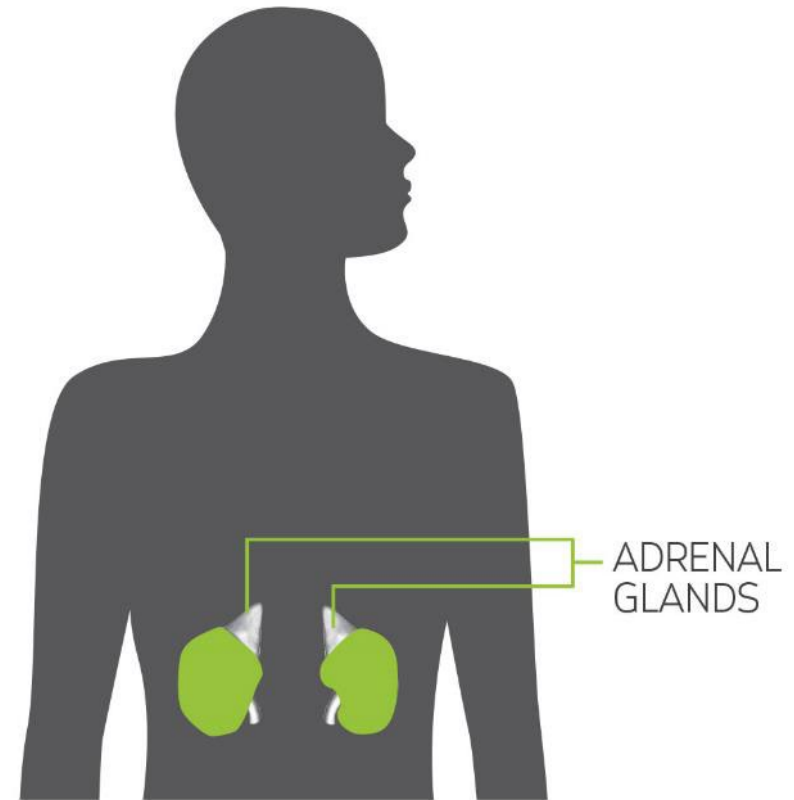
©AMEND

# Adrenocortical Cancer (ACC)

- Cancer of the outer layer of the adrenal gland (cortex)
- Usually found incidentally by scans for other reasons
- Over produces steroid hormones such as cortisol causing symptoms
- Extremely rare

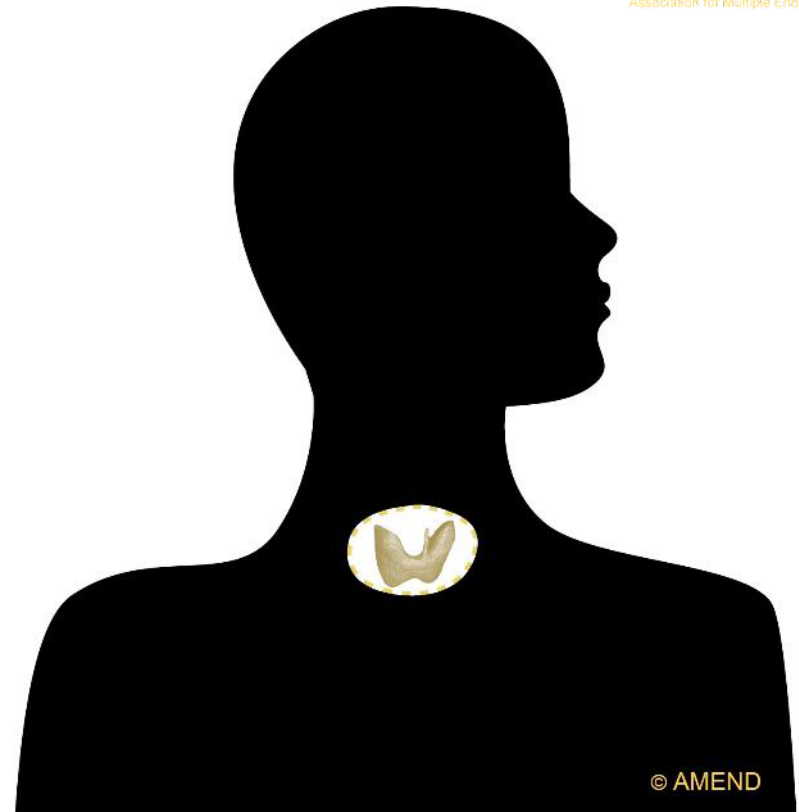
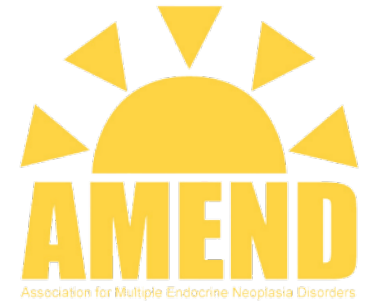


<https://accsupport.org.uk/>



# Medullary Thyroid Cancer (MTC)

- Medullary thyroid cancer that is not inherited (sporadic)
- rare



© AMEND

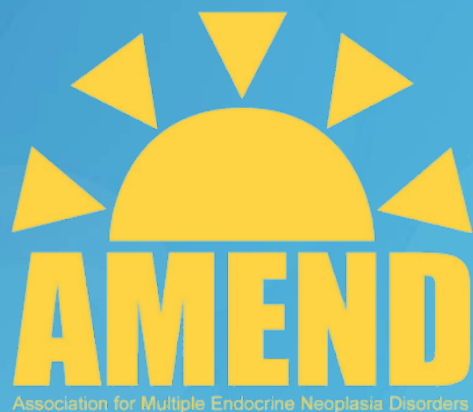
<https://www.amend.org.uk/patients/information/mtc/>

# Our Unmet Needs

- Late diagnosis
- Lack of psychological support from diagnosis
- Lack of research into our diseases:
  - Problem for gaining evidence to be granted access to new therapies and to informing best practice through Guidelines
- Lack of collaborative international research







# 20 Years of AMEND



# How it all began

## **Do you have MEN1?**

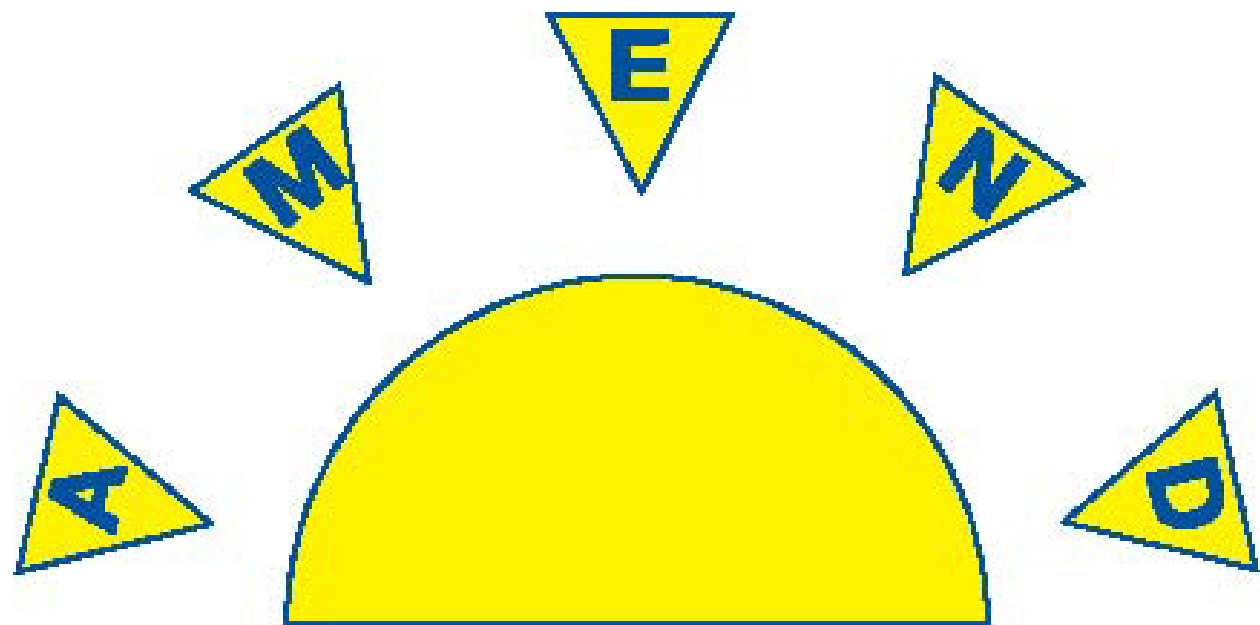
**My daughter and I have been diagnosed with multiple endocrine neoplasia type 1 and are looking for others to potentially set up a UK patient group. If you are interested, please call Liz on 01785 555666**



Liz Dent and Emily Fazal



The Very  
First  
Information  
Day, 2003



**Association for**  
**MULTIPLE**  
**ENDOCRINE**  
**NEOPLASIA**  
**Disorders**

2003  
Registered  
as a Charity  
in England  
& Wales

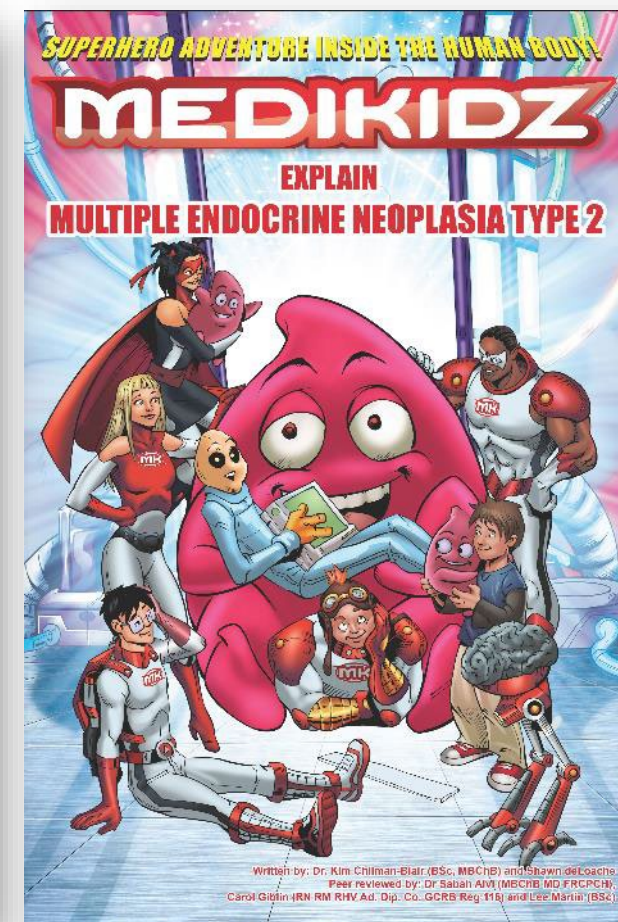
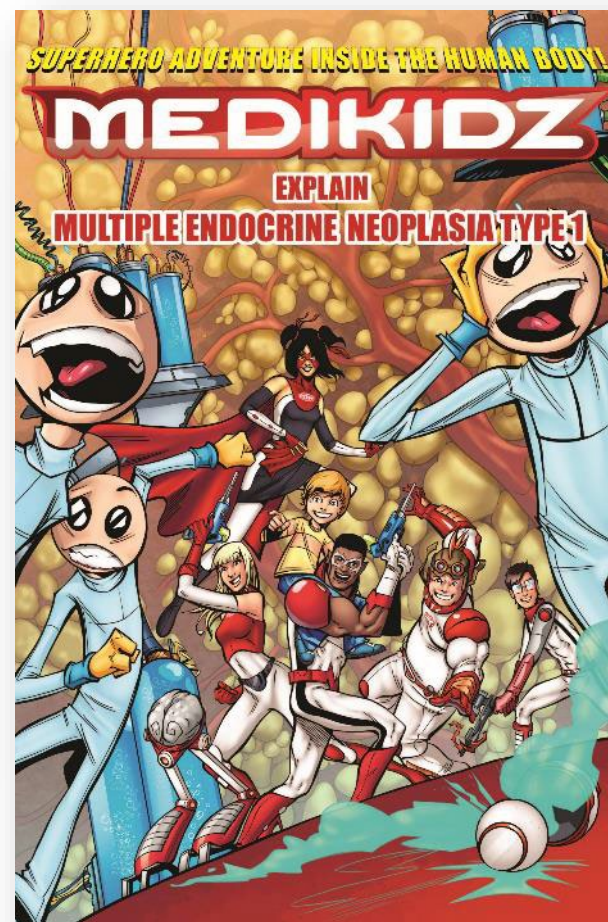
# Launch of the MEN Passports (2009)



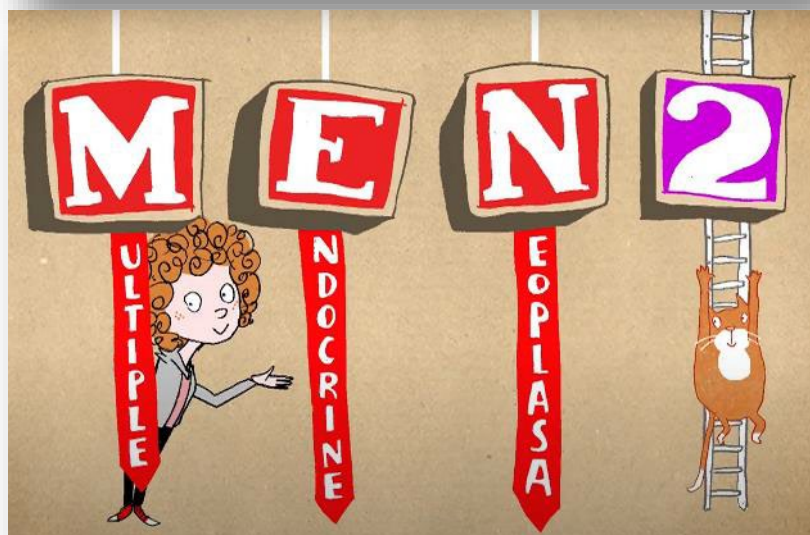
# Gaining recognition from medical societies and a new look!



# Resources – for children (2012)



# Resources – for children (2012)

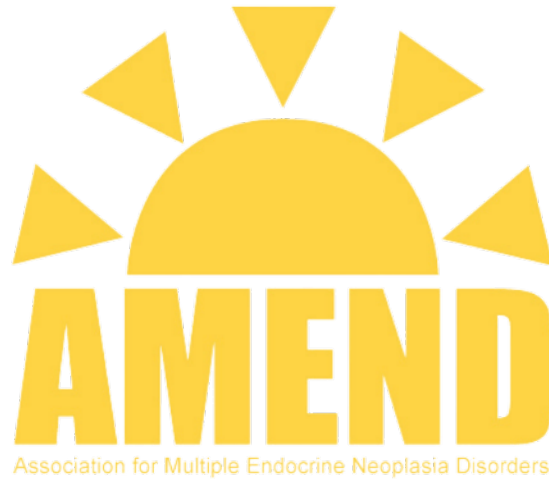




# Widened Remit



[www.accsupport.org.uk](http://www.accsupport.org.uk)

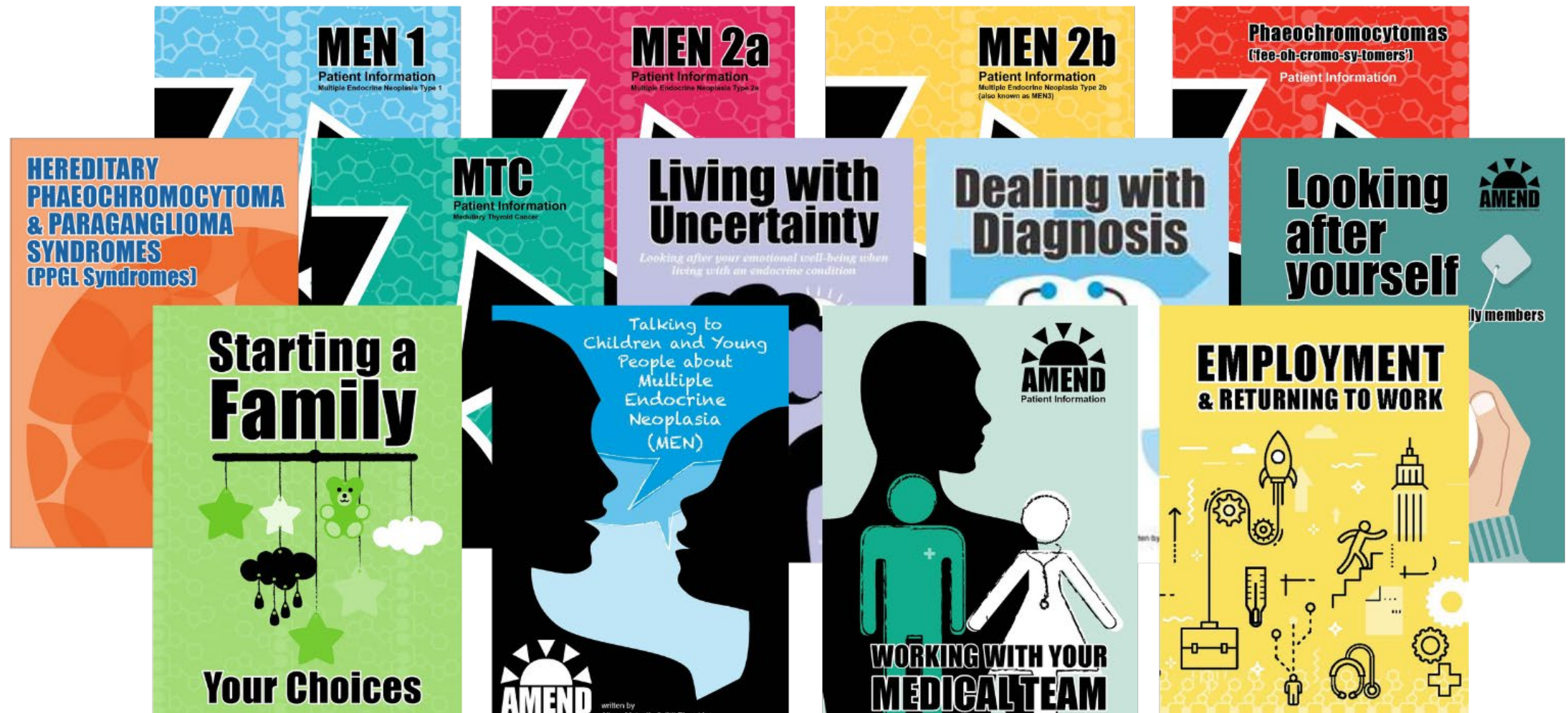


[www.amend.org.uk](http://www.amend.org.uk)









[www.phaeoparasupport.org.uk](http://www.phaeoparasupport.org.uk)

# Resources - booklets



# Resources - films



|  |   |  |
|--|---|--|
| <p>400TM 46 400TM</p> <p><b>MEN1</b></p>  <p>A close-up shot of a bald man in a dark jacket and a woman with long dark hair embracing each other. The man is smiling and looking towards the woman.</p>                                 | <p>47 400TM 48</p> <p><b>MEN2</b></p>  <p>A person in a red jacket stands on a wooden railing, looking out over a vast green valley with rolling hills and a small town in the distance under a blue sky.</p> | <p>400TM 49 400TM</p> <p><b>MEN3</b></p>  <p>A baby is crawling on a colorful, geometric-patterned rug, playing with several toys including a yellow ball and a wooden toy.</p>   |
| <p>400TM 46 400TM</p> <p><b>PPGL</b></p> <p>Introducing SDH Syndromes</p> <hr/> <p>DIAGNOSIS</p>  <p>A close-up of a woman's face, partially obscured by colorful, semi-transparent overlays in shades of purple, blue, and green.</p> | <p>47 400TM 48</p> <p><b>ACC</b></p> <p>Talking about ACC</p> <p>Living with ACC</p>  <p>A close-up of a heart-shaped metal ornament with intricate designs, set against a dark background.</p>              | <p>400TM 49 400TM</p> <p><b>YOUNGSTERS</b></p> <p>Young people chat about the aspects of</p> <p><b>living with MEN</b></p>  <p>A group of young people are sitting together, smiling and talking. The scene is brightly lit and appears to be an indoor setting.</p> |

# New Phaeo Para Resource for kids



 **Saskia Grassie**  
@SaskiaGrassie

Replying to @ROBYOUNG @PhaeoU and 2 others

Congratulations! 🎉 It's a beautiful little film and I immediately wanted to find out more about [#PGLSyndromes](#) and the importance of early detection. Having access to engaging resources which help open discussion with children about difficult and sensitive topics so valuable

08:07 · 18/11/2022 · [Twitter for iPhone](#)

**Me, My Mum, and Dave**

...and so much more





2012 - 2022

**£170,000**

RESEARCH  
GRANTS

## Research

- MEN1
  - MEN2
  - MEN3
  - PPGL
  - ACC
  - MTC
- 
- x2 PHD projects regarding MEN3

# Volunteers are our lifeblood

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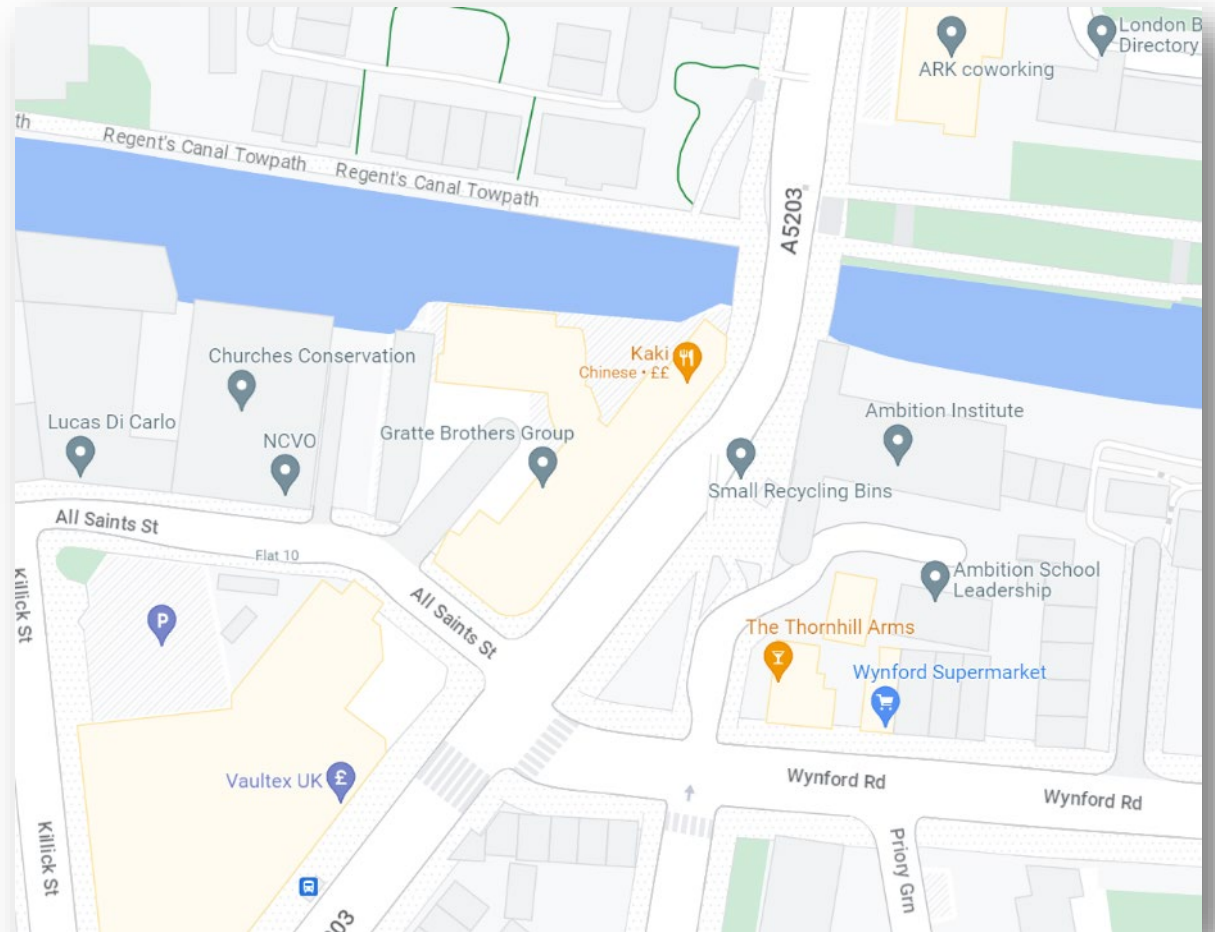
- Trustees
- Medical Advisory Teams
- Peer Support Volunteer Team
- Fundraisers
- Donors
- Grantees
- APID speakers
- And many, many more!



# Thank You!



- Join us in the Thornhill Arms pub across the road from 6pm to celebrate 20 years of AMEND!





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