Insight Report: Molecular Phenomics Patient Diagnosis Online Survey

Insight report summarising the findings from an online public involvement survey to explore commonly prone to missed, late or incorrectly diagnosed conditions/diseases across patients to support the development of the Molecular Phenomics Theme.

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Background

The National Institute for Health Research (NIHR) currently funds 20 Biomedical Research Centres (BRCs) across England. These are collaborations between world-leading universities and NHS organisations that bring together academics and clinicians to translate lab-based scientific breakthroughs into potential new treatments, diagnostics, and medical technologies. The Imperial BRC is a collaboration between Imperial College, London and Imperial College Healthcare NHS Trust and is currently funded until 2022. It has 12 research themes, 4 of which are cross cutting.

As part of the reapplication for the BRC competition run by the NIHR, a public involvement online survey was conducted to assist with the development of the Molecular Phenomics Theme and its programme of work aiming to improve diagnostic testing. The survey was facilitated by the Patient Experience Research Centre (PERC), a core facility of the current Imperial BRC and aimed to capture conditions and diseases which were prone to late, missed or incorrect diagnoses.

Approach and purpose

Public involvement was considered a crucial component of the development of this proposed theme's programme of research. Through the online survey the Molecular Phenomics Theme particularly wanted to understand from the perspective of patients and the public which conditions/diseases are the most difficult to diagnose with the aim to use the information to help develop diagnostic tests to speed up diagnosis for these medical conditions in the future.

Efforts were made to disseminate the survey to those who had not previously taken part in public involvement in research activities at Imperial College, London, or at all. The importance of capturing these views was to increase the representativeness of those individuals whose voices are not usually heard in public involvement in research which is a continuing area of focus.

Survey format

The survey was hosted on Qualtrics, an online survey platform. The survey included 3 questions, which asked respondents to relay any misdiagnoses they had received, the accurate diagnosis they had now received and the approximate length of time it took to be accurately diagnosed. The survey also captured the demographics of respondents (age, gender and ethnicity).

Survey dissemination

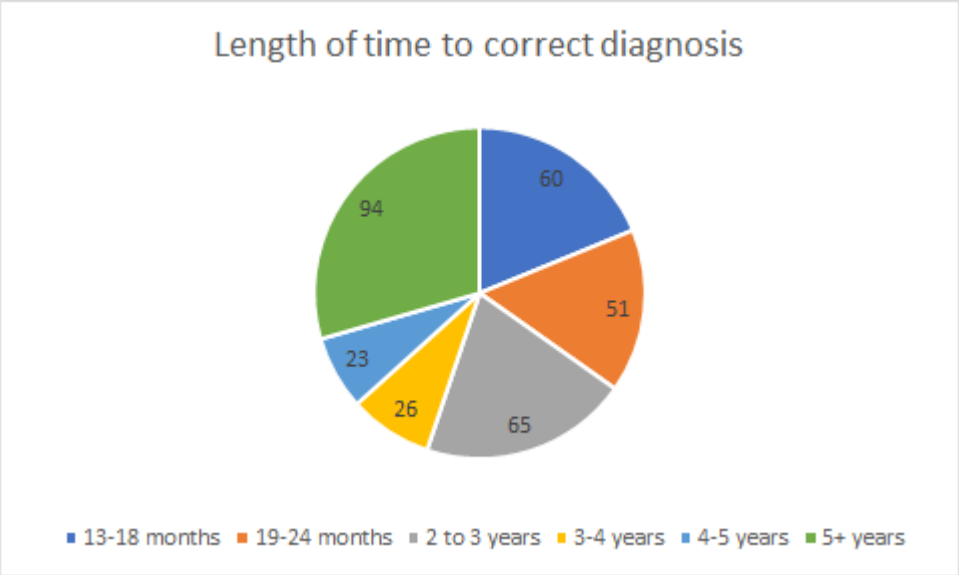
The survey was disseminated online between August – September 2021 through the following routes:

- By the North West London Clinical Commissioning Group (CCG) Engagement Manager posting it on the online "Nextdoor" platform
- Through the VOICE global online platform
- To existing PERC networks (PERC mailing list, Imperial Young Persons Advisory Network and through the PERC team)
- By asking members of the Imperial BRC Public Advisory Panel to disseminate it to relevant individuals across their wider community
- Through the CHARIOT Research Registry
- Through Twitter

Key Insight Summary

In total, 1023 responses (see Appendix 1 for demographics of respondents) were received from individuals across the UK using the online survey platform (Qualtrics). Individuals were invited to complete the survey if they had received a missed, late, or incorrect diagnosis.

A preliminary analysis was conducted which considered only respondents who had reported receiving a clear and accurate final diagnosis after a period of one or more years of misdiagnosis (n=319; broken down in the figure below), representing the most severe (in terms of both personal and healthcare system-related impact) cases polled:



The data reveals previously unseen insight into both diseases which are difficult to accurately diagnose and diseases that masquerade as others, resulting in misdiagnoses and mistreatment. A more comprehensive analysis is ongoing, however, the following diseases were of initial interest due to their frequent reporting:

- Cancer was the most widely reported final diagnosis (n=19), including bowel, bladder, kidney, prostate, cervix, skin and breast.
- Myalgic Encephalomyelitis/Chronic Fatigue Syndrome (ME/CFS) was reported seven times, mistaken for depression.
- Rheumatoid Arthritis was reported seven times, mistaken for wear & tear, bone fractures and Raynaud's.
- Parkinson's disease was reported 6 times, interestingly with no misdiagnoses in any reported cases, but all cases requiring between 1 and 3 years to diagnose.
- Hiatus hernia was reported 6 times, mistaken for a wide range of conditions including COPD, bowel cancer, IBS, and chronic cough.
- As a pair of interest, chronic obstructive pulmonary disease (COPD) was reported 5 times, mistaken for asthma. Whereas asthma was reported 6 times with 5 of 6 patients reporting more than 2 years. It is important to note that the treatment for these diseases is different, with asthma requiring suppression of chronic inflammation and COPD requiring management of symptoms.
- Fibromyalgia was reported 5 times.
- Hyperthyroidism was reported 5 times, most frequently mistaken for depression.

- Adult growth hormone deficiency was reported twice, both times with time to accurate diagnosis exceeding 5 years. It was mistaken for mental health disorders, malingering, depression, and anxiety.
- Bipolar disorder was reported 5 times, mistaken for depression, anxiety, schizophrenia. 5/5 patients reported at least 3 years to diagnosis.
- Bronchiectasis was reported 5 times, generally mistaken for asthma, acid reflux, lung infection or viral illness.
- Endometriosis was reported 4 times, most frequently mistaken for IBS. Crohn's was reported 3 times, also mistaken for IBS (as well as lactose intolerance). Chronic pancreatitis and Coeliac were both mistaken for IBS. Both bowel and ovarian cancer were misdiagnosed as IBS. Interestingly, IBS, in the absence of hiatus hernia, was not reported.

The above preliminary findings identify the need for improved disease stratification (classification into different groups), specifically where symptom reporting may be general (e.g. where many distinct diseases appear to be mistaken for IBD). Further, the heterogeneity (diversity) in undiagnosed diseases (with the majority of reports appearing as single cases in the collated data) represents a substantial challenge for accurate disease diagnosis and future treatment. Powerful metabolic phenotyping technologies are ready to be able to assist here, if the field can be moved onward from exploring the metabolic underpinnings of single diseases (vs. healthy controls) to looking across diseases that are prone to misdiagnosis or that fall under the umbrella of a single symptom set.

How the insights were used

The insight report summarising the key findings from the online survey was made available to the Theme leads and the BRC Executive in order to shape the BRC application and for future research development in this area. A full report on all public involvement activities undertaken in preparation for the BRC application can be found here.

Those who completed the survey were also given the opportunity to sign up for future public involvement, engagement, and participation opportunities.

We would like to thank all those members of the public who gave their time and thoughtful insights through these activities, and the researchers who engaged enthusiastically in the process.

Appendix 1: Demographics of Respondents

Characteristics	n (%)
Age (in years)	
Mean (range)	70 (13 – 93)
Age groups (in years)	
Under 18	1 (0.1)
18-24	3 (0.29)
25-34	11 (1.08)
35-44	15 (1.47)
45-54	35 (3.42)
55-64	122 (11.93)
65-74	489 (47.8)
75 - 84	279 (27.27)
85 +	36 (3.52)
Not provided	32 (3.13)
Gender	
Female	606 (59.24)
Male	400 (39.1)
Non-binary/Gender Variant	1 (0.1)
Transwoman	1 (0.1)
Prefer not to say /Not provided	18 (1.76)
Ethnic group	
White	
English/Welsh/Scottish/Northern Irish/British	783 (76.54)
Irish	22 (2.15)
Gypsy or Irish Traveller	0 (0.0)
Other White background	106 (10.36)
Mixed/Multiple Ethnicity	
White and Black African	0 (0.0)
White and Black Caribbean	3 (0.29)
White and Asian	3 (0.29)
Other Mixed/Multiple background	6 (0.59)
Asian/Asian British	
Indian	34 (3.32)
Pakistani	1 (0.1)
Bangladeshi	2 (0.2)
Chinese	5 (0.49)
Other Asian background	9 (0.88)
Black/African/Caribbean/Black British	
Black	1 (0.1)
African	2 (0.2)
Caribbean	5 (0.49)
Other Black/African/Caribbean background	1 (0.1)

Other	
Arab	4 (0.39)
Any other ethnic group	16 (1.56)
Prefer not to say/not provided	23 (2.25)