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RareDx: A national training event to raise awareness of rare diseases in primary care

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Introduction

Rare diseases are a major investigative area of our time and research in the field continues to make great strides. Until now, it has been reserved as an specialist area however, with interest, knowing and technological capabilities growing, it is starting to enter the sphere of primary care [1]. The 100,000 genome project has been a groundbreaking study that aims to sequence genomes from approximately 70,000 patients who suffer from rare diseases [2]. It is hoped that this will kick-start the understanding of diseases within the UK Genomics industry, as well as provide vital breakthroughs in our understanding of their pathophysiology. The knock-on effects from this study are numerous; having an awareness of the diagnostic challenges and how to interpret common tests can help with early identification and management of these rare and disabling conditions.

Aims

To deliver a national training event aimed at General Practitioners, to raise the awareness and understanding of rare diseases

Secondary Aims:

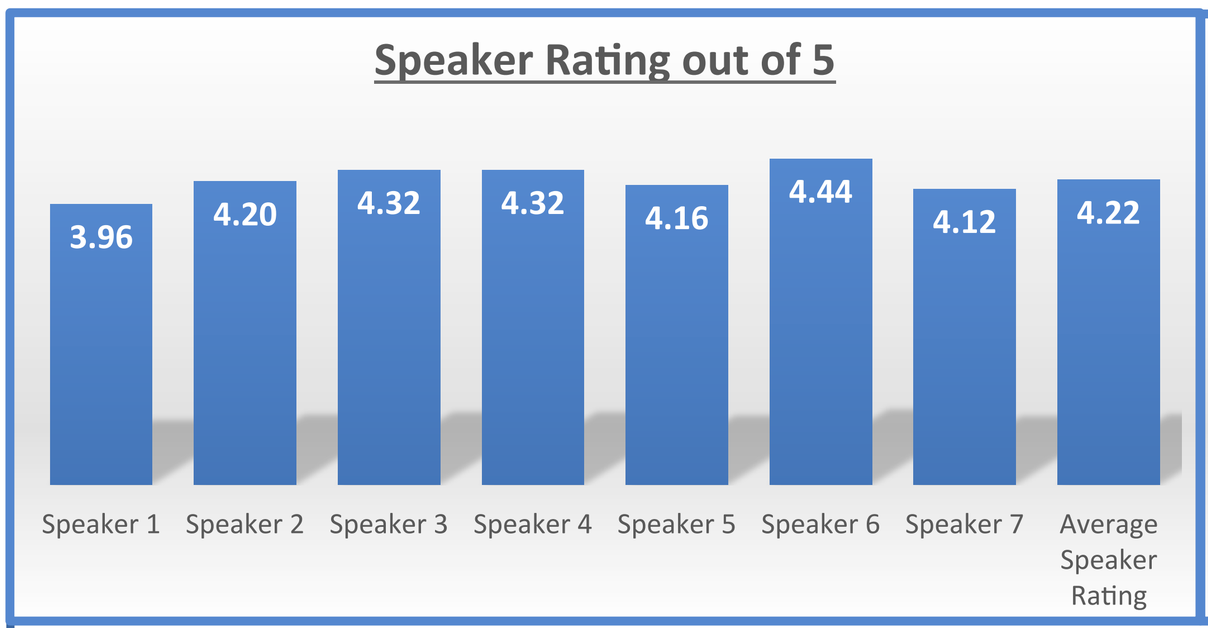
To understand and appreciate:

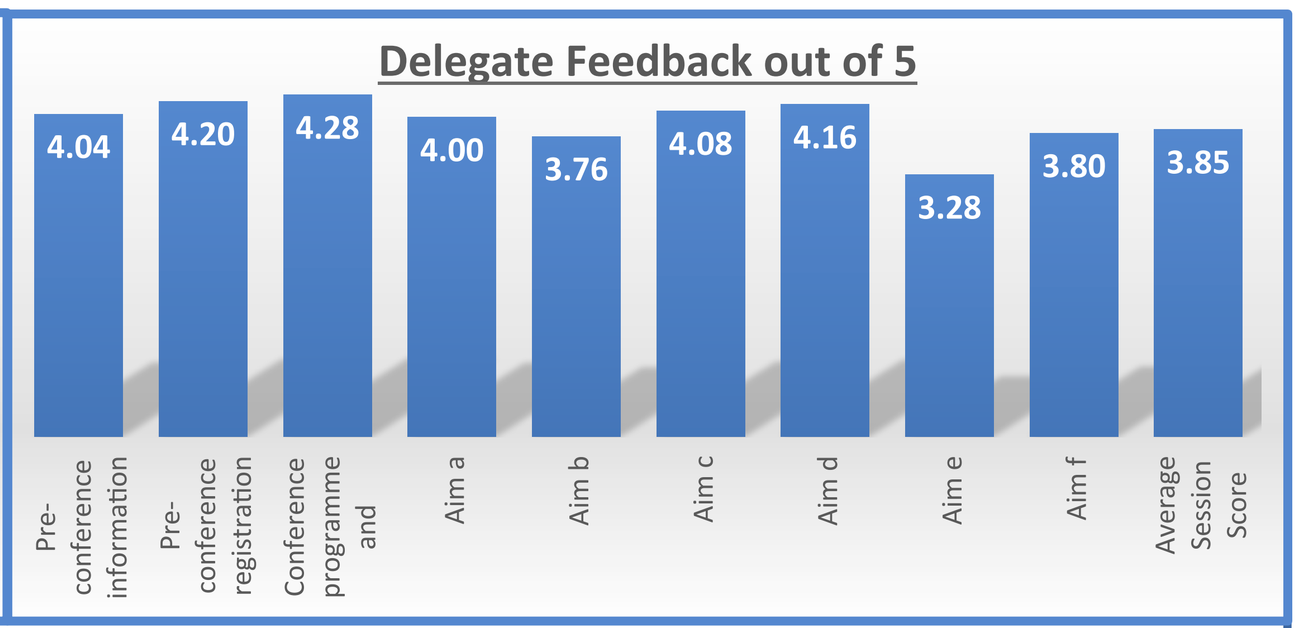
a. Diagnostic challenges involved in rare diseases e.g. due to unusual constellations of presentations	b. The correct pathway to recruit for the 100,000 genome project	c. The state of research in Rare Diseases and how primary care can contribute	d. Importance of rare disease knowledge in primary care	e. Interpretation of common genetic tests	f. The primary management of rare diseases in childhood

Methods

A committee consisting of 4 medical students was formed in order to facilitate the organisation of the day. Furthermore, the team liaised with a team at Queen Elizabeth Hospital, Birmingham (QEHB) to arrange the location for the event. Approval from the Royal College of Physicians was sought in order to provide CPD accreditation for this event. After speaking to a number of fellow healthcare professionals and future students, we ascertained that there were a number of areas that we wanted to address over the course of the day. Our speakers were chosen for their breadth and depth of knowledge in the field and we tailored the content to match our speakers' areas of expertise. The course was provided for free thanks to sponsorship sought from Alexion Inc. and Sanofi Genzyme.

Following the event, participants were asked to complete an online feedback form, which contained a Likert scale (from 1-5, 1 = poor, 5 = excellent) to rate their thoughts on the quality of the content provided by the speakers and understanding how well the event's aims were met [3]. An online form was chosen as this made it easier for the delegates to respond and provided a simpler method of analysing the ratings. Donations to Birmingham Children's Hospital were encouraged in order to support Rare Diseases Research. Results were subsequently collated on Microsoft Excel and analysed appropriately.





Results and Discussion

Overall, we are delighted with the feedback given across the board, especially considering this event is the first of it's kind. Of the 84 participants registered as attending, 26 attended. Feedback response rate was 96%. We enabled this by providing certificates of attendance in exchange for receipt of feedback. Speakers were rated at 'Very Good', with an average rating of 4.22 (3.96-4.44). Participants felt the publicity and registration process were also 'Very Good', with an average rating of 4.04 and 4.20 respectively. In general, the aims outlined at the beginning of the course were met, with sessions rated as either 'Good' or 'Very Good'.

Clearly, despite the excellent feedback for the event, there is scope for improvement, particularly in helping practitioners understand the recruiting pathway for the 100,000 genome project and the interpretation for common genomic tests. These formed some of our poorest performing categories and it is imperative that we remedy the future talks delivered in these areas in order to meet the core aims and aspirations of the Rare Diseases Event. We are delighted to have put rare diseases at the forefront of thought, and look forward to making future events even better.

References

1. The UK Strategy for Rare Diseases [Internet]. London: Department of Health; 2013 [cited 13 April 2018]. Available from: https://assets.publishing.service.gov.uk/government/uploads/system/uploads/attachment_data/file/260562/UK_Strategy_for_Rare_Diseases.pdf
2. The 100,000 Genomes Project | Genomics England [Internet]. Genomics England. 2018 [cited 13 April 2018]. Available from: https://www.genomicsengland.co.uk/the-100000-genomes-project/
3. McLeod S. Likert Scale | Simply Psychology [Internet]. Simplypsychology.org. 2018 [cited 13 April 2018]. Available from: https://www.simplypsychology.org/likert-scale.html