Making progress with newborn screening

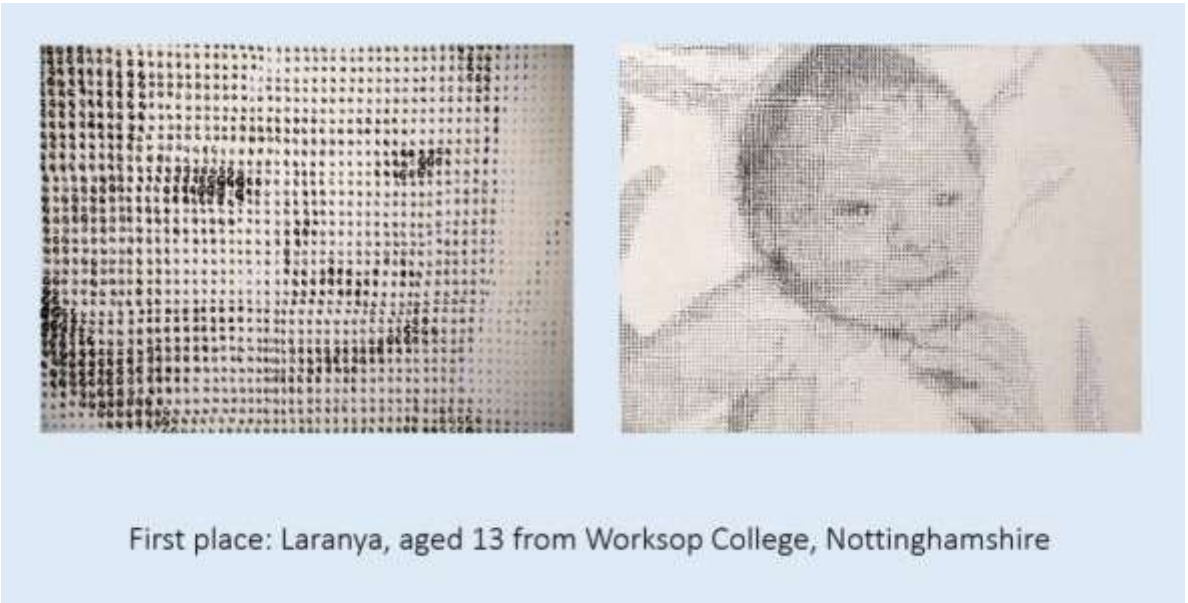
St Anne's College, Oxford • 25th February 2025 • 9:30 – 18:15

In this event, we hope to explore some of the challenges around balancing the benefits of extending screening for babies with rare conditions, with the involvement this requires from babies who do not stand to personally benefit.

We hope that this will be a forum for constructive and pragmatic discussion, looking at where things currently stand with newborn screening, and considering how we can make the most of current research opportunities to test and improve the screening process.

This event is organised by the Centre for Personalised Medicine with support from Cambridge Prisms.

(The art throughout this programme was created by young people in Years 7 to 9 for the 2023-24 Centre for Personalised Medicine Youth Art Competition, on the theme of screening newborn babies for disease.)



First place: Laranya, aged 13 from Worksop College, Nottinghamshire

Agenda

9:30	**Registration & coffee**	
10:00	*Newborn screening – the case for widening it, and the hurdles that we need to anticipate*	Professor Anneke Lucassen, Director, Centre for Personalised Medicine

Focus 1: what would success look like in the context of a rare disease screening programme?

This session will look at qualities of successful screening programmes, and consider how principles around screening should account for very rare conditions, where we cannot expect to have a wealth of data to guide decisions.

Chair: Professor Anneke Lucassen

10:15	*How to make good public policy screening recommendations for rare diseases: benefits, harms, opportunity costs*	Professor Anne Mackie, Director of Screening, Public Health England
10:30	*Do the current screening criteria in the UK set very rare diseases up to fail?*	Nick Meade, Director of Policy, Genetic Alliance UK
10:45	*Principles for including conditions in the Generation Study*	Dr Ellen Thomas, Chief Medical Officer, Genomics England
11:00	*Building a health economic case for extending newborn screening*	Dr James Buchanan, Senior Lecturer, Health Economics and Policy Research Unit, Queen Mary University of London
11:15	*Panel discussion – how should screening recommendations take account of rare disease?*	Professor Anne Mackie Nick Meade Dr Ellen Thomas Dr James Buchanan Professor Sian Taylor-Phillips, Professor of Population Health, University of Warwick
12:15	*Cambridge Prisms: Precision Medicine*	Professor Dame Anna Dominiczak, Editor-in-Chief, Cambridge Prisms: Precision Medicine

12:30 **Lunch**

Highly commended: Ahona, aged 13 from Francis Holland School, London

Focus 2: navigating new uncertainties

This session will discuss the challenges of interpreting genomic variation in a screening context, the challenges for families and clinicians of managing uncertainty, and the challenges of living with a rare condition.

Chair: Dr Susie Weller

13:30	*The scientific challenge of predicting phenotype from genotype*	Professor Caroline Wright, Clinical and Biomedical Sciences, University of Exeter
13:45	*Challenge of managing uncertainty when incorporating expanded genetic screening into CF Newborn Bloodspot Screening*	Dr Jane Chudleigh, King's College London
14:00	*The challenges of living with a rare disease diagnosis*	Dr Sarah Wynn, Chief Executive Officer, Unique
14:15	*The challenge of providing clinical care in situations of uncertainty*	Dr Robin Lachmann, Consultant in Inherited Metabolic Disease, National Hospital for Neurology and Neurosurgery, Queen Square, London
14:30	*Panel discussion – how should we prepare parents and the NHS for navigating new uncertainties?*	Professor Caroline Wright Dr Jane Chudleigh Dr Sarah Wynn Dr Robin Lachmann Dr Judith Hayward, RCGP joint clinical representative in Genomic Medicine Dr Jonathan Roberts, Research Genetic Counsellor, The Synapse Centre for Neurodevelopment
15:30	**Coffee break**	

Runner-up: Scarlett, aged 12 from Worksop College, Nottinghamshire

Focus 3: the opportunities and questions raised by screening using genomes as opposed to more targeted options

This session will explore the potential benefits of research with genomic data, and the complexities of asking consent for genomic data collection from healthy babies.

Chair: Professor Caroline Wright

16:00	*Session introduction*	Professor Caroline Wright
16:05	*Publics views about contributing genomic data for research*	Dr Richard Milne, Head of Research and Dialogue, Engagement and Society, Wellcome Sanger Institute
16:20	*Ethical considerations for projects involving 'healthy' babies*	Professor Dominic Wilkinson, Director of Medical Ethics, Oxford Uehiro Centre for Practical Ethics
16:35	*Finding responsible ways to let innovation flourish*	Professor Michael Parker, Director of the Ethox Centre, University of Oxford
16:50	*Panel discussion – how can newborn genome studies support parents to engage with what it means to contribute genomic data, as well as what it means to have expanded screening?*	Dr Richard Milne Professor Dominic Wilkinson Professor Michael Parker Dr Meekai To, Clinical Fellow in Genomics, Genomics England Dr Celine Lewis, Principal Research Fellow in Genomics, UCL Great Ormond Street Institute of Child Health
17:30	*Closing remarks/common ground*	Professor Anneke Lucassen
	Visual minutes	Zuhura Plummer
17:40	**Drinks reception until 18:15**	

Highly commended: Parampreet, aged 11 from Higham Lane School, Warwickshire

Highly commended: Rayan, aged 13 from Oaklands Secondary School, London