

<https://doi.org/10.1038/s41467-019-10161-9>

OPEN

Author Correction: CHD3 helicase domain mutations cause a neurodevelopmental syndrome with macrocephaly and impaired speech and language

Lot Snijders Blok, Justine Rousseau, Joanna Twist et al.[#]

Correction to: *Nature Communications* <https://doi.org/10.1038/s41467-018-06014-6>, published online 05 November 2018.

The HTML and PDF versions of this Article were updated after publication to remove images of one individual from Fig. 1.

Published online: 02 May 2019



Open Access This article is licensed under a Creative Commons Attribution 4.0 International License, which permits use, sharing, adaptation, distribution and reproduction in any medium or format, as long as you give appropriate credit to the original author(s) and the source, provide a link to the Creative Commons license, and indicate if changes were made. The images or other third party material in this article are included in the article's Creative Commons license, unless indicated otherwise in a credit line to the material. If material is not included in the article's Creative Commons license and your intended use is not permitted by statutory regulation or exceeds the permitted use, you will need to obtain permission directly from the copyright holder. To view a copy of this license, visit <http://creativecommons.org/licenses/by/4.0/>.

© The Author(s) 2019

Lot Snijders Blok^{1,2,3}, Justine Rousseau⁴, Joanna Twist⁵, Sophie Ehresmann⁴, Motoki Takaku⁵, Hanka Venselaar⁶, Lance H. Rodan⁷, Catherine B. Nowak⁷, Jessica Douglas⁷, Kathryn J. Swoboda⁸, Marcie A. Steeves⁹, Inderneel Sahai⁹, Connie T.R.M. Stumpel¹⁰, Alexander P.A. Stegmann¹⁰, Patricia Wheeler¹¹, Marcia Willing¹², Elise Fiala¹², Aaina Kochhar¹³, William T. Gibson^{14,15}, Ana S.A. Cohen^{14,15}, Ruky Agbahovbe^{14,15}, A. Micheil Innes¹⁶, P.Y. Billie Au¹⁶, Julia Rankin¹⁷, Ilse J. Anderson¹⁸, Steven A. Skinner¹⁹, Raymond J. Louie¹⁹, Hannah E. Warren¹⁹, Alexandra Afenjar²⁰, Boris Keren^{21,22}, Caroline Nava^{21,22,23}, Julien Buratti²¹, Arnaud Isapof²⁴, Diana Rodriguez²⁵, Raymond Lewandowski²⁶, Jennifer Propst²⁶, Ton van Essen²⁷, Murim Choi²⁸, Sangmoon Lee²⁸, Jong H. Chae²⁹, Susan Price³⁰, Rhonda E. Schnur³¹, Ganka Douglas³¹, Ingrid M. Wentzensen³¹, Christiane Zweier³², André Reis³², Martin G. Bialer³³, Christine Moore³³, Marije Koopmans³⁴, Eva H. Brilstra³⁴, Glen R. Monroe³⁴, Koen L.I. van Gassen³⁴, Ellen van Binsbergen³⁴, Ruth Newbury-Ecob³⁵, Lucy Bownass³⁵, Ingrid Bader³⁶, Johannes A. Mayr³⁷, Saskia B. Wortmann^{37,38,39}, Kathy J. Jakielski⁴⁰, Edythe A. Strand⁴¹, Katja Kloth⁴²,

Correspondence and requests for materials should be addressed to S.E.F. (email: simon.fisher@mpi.nl) or to P.M.C. (email: p.campeau@umontreal.ca). [#]A full list of authors and their affiliations appears at the end of the paper.

Tatjana Bierhals⁴², The DDD study, John D. Roberts⁵, Robert M. Petrovich⁵, Shinichi Machida⁴³, Hitoshi Kurumizaka⁴³, Stefan Lelieveld¹, Rolph Pfundt¹, Sandra Jansen^{1,3}, Pelagia Deriziotis², Laurence Faivre^{44,45}, Julien Thevenon^{44,45}, Mirna Assoum^{44,45}, Lawrence Shriberg⁴⁶, Tjitske Kleefstra^{1,3}, Han G. Brunner^{1,3,10}, Paul A. Wade⁵, Simon E. Fisher^{2,3} & Philippe M. Campeau^{4,47}

¹Department of Human Genetics, Radboud University Medical Center, Nijmegen 6500HB, The Netherlands. ²Language and Genetics Department, Max Planck Institute for Psycholinguistics, Nijmegen 6500AH, The Netherlands. ³Donders Institute for Brain, Cognition and Behaviour, Radboud University, Nijmegen 6500HE, The Netherlands. ⁴CHU Sainte-Justine Research Center, Montreal QC H3T 1C5, Canada. ⁵National Institute of Environmental Health Sciences, Research Triangle Park NC 27709, USA. ⁶Centre for Molecular and Biomolecular Informatics, Radboud Institute for Molecular Life Sciences, Radboud University Medical Center, Nijmegen 6500HB, The Netherlands. ⁷Division of Genetics and Genomics, Boston Children's Hospital, Boston MA 02115, USA. ⁸Department of Neurology, Massachusetts General Hospital and Harvard Medical School, Boston MA 02114, USA. ⁹Department of Medical Genetics, Massachusetts General Hospital, Boston MA 02114, USA. ¹⁰Department of Clinical Genetics and GROW-School for Oncology and Developmental Biology, Maastricht University Medical Center, Maastricht 6202AZ, The Netherlands. ¹¹Nemours Children's Clinic, Orlando FL 32827, USA. ¹²Division of Genetics and Genomic Medicine, Department of Pediatrics, Washington University School of Medicine, St. Louis MO 63110, USA. ¹³Valley Children's Hospital, Madera CA 93636, USA. ¹⁴British Columbia Children's Hospital Research Institute, Vancouver BC V5Z 4H4, Canada. ¹⁵Department of Medical Genetics, University of British Columbia, Vancouver BC V6H 3N1, Canada. ¹⁶Department of Medical Genetics and Alberta Children's Hospital Research Institute, Cumming School of Medicine, University of Calgary, Calgary AB T2N 4N1, Canada. ¹⁷Department of Clinical Genetics, Royal Devon and Exeter NHS Foundation Trust (Heavitree), Exeter EX2 5DW, UK. ¹⁸Division of Genetics, Department of Medicine, University of Tennessee Medical Center, Knoxville TN 37920, USA. ¹⁹Greenwood Genetic Center, Greenwood SC 29646, USA. ²⁰GRC ConCer-LD, Sorbonne Universités, UPMC Univ Paris ; Department of Medical Genetics and Centre de Référence Malformations et maladies congénitales du cervelet et déficiences intellectuelles de causes rares, Armand Trousseau Hospital, GHUEP, AP-HP, Paris 75012, France. ²¹AP-HP, Hôpital de la Pitié-Salpêtrière, Département de Génétique, Paris 75013, France. ²²Groupe de Recherche Clinique (GRC) 'déficience intellectuelle et autisme' UPMC, Paris 75005, France. ²³INSERM, U 1127, CNRS UMR 7225, Institut du Cerveau et de la Moelle épinière, ICM, Sorbonne Universités, UPMC Univ Paris 06 UMR S 1127, 75013 Paris, France. ²⁴GRC ConCer-LD, Sorbonne Universités, UPMC Univ Paris 06; Department Child Neurology and Reference Center for Neuromuscular Diseases "Nord/Est/Ile-de-France", FILNEMUS, Armand Trousseau Hospital, GHUEP, AP-HP, Paris 75012, France. ²⁵GRC ConCer-LD, Sorbonne Universités, UPMC Univ Paris 06; Department of Child Neurology and National Reference Center for Neurogenetic Disorders, Armand Trousseau Hospital, GHUEP, AP-HP, INSERM U1141, 75012 Paris, France. ²⁶Clinical Genetics Division, Virginia Commonwealth University Health System, Richmond VA 23298, USA. ²⁷Clinical Genetics Department, University Medical Center Groningen, Groningen 9700RB, The Netherlands. ²⁸Department of Biomedical Sciences, Seoul National University College of Medicine, Seoul 08826, Republic of Korea. ²⁹Department of Pediatrics, Seoul National University College of Medicine, Seoul National University Children's Hospital, Seoul 08826, Republic of Korea. ³⁰Oxford University Hospitals NHS Foundation Trust, Oxford OX3 7HE, UK. ³¹GeneDx, Gaithersburg MD 20877, USA. ³²Institute of Human Genetics, Friedrich-Alexander-Universität Erlangen-Nürnberg, Erlangen 91054, Germany. ³³Northwell Health, Division of Medical Genetics and Genomics, Great Neck NY 11021, USA. ³⁴Department of Genetics, University Medical Center Utrecht, Utrecht University, Utrecht 3508AB, The Netherlands. ³⁵University Hospitals Bristol, Department of Clinical Genetics, St Michael's Hospital, Bristol BS2 8EG, UK. ³⁶Department of Clinical Genetics, University Children's Hospital, Paracelsus Medical University, Salzburg A-5020, Austria. ³⁷Department of Pediatrics, Salzburger Landeskliniken and Paracelsus Medical University, Salzburg A-5020, Austria. ³⁸Institute of Human Genetics, Technische Universität München, Munich 81675, Germany. ³⁹Institute of Human Genetics, Helmholtz Zentrum München, Neuherberg 85764, Germany. ⁴⁰Communication Sciences and Disorders, Augustana College, Rock Island IL 61201, USA. ⁴¹Department of Neurology, Mayo Clinic, Rochester MN 55905, USA. ⁴²Institute of Human Genetics, University Medical Center Hamburg-Eppendorf, Hamburg 20246, Germany. ⁴³Waseda University, Tokyo 169-8050, Japan. ⁴⁴Equipe Génétique des Anomalies du Développement, Université de Bourgogne-Franche Comté, Dijon 21070, France. ⁴⁵Centre de Génétique et Centre de Référence Anomalies du Développement et Syndromes Malformatifs, FHU TRANSLAD, Hôpital d'Enfants, CHU Dijon et Université de Bourgogne, Dijon 21079, France. ⁴⁶Waisman Center, Phonology Project, Madison WI 53705-2280, USA. ⁴⁷Sainte-Justine Hospital, University of Montreal, Montreal QC H3T 1C5, Canada. These authors contributed equally: Lot Snijders Blok, Justine Rousseau, Joanna Twist. These authors jointly supervised this work: Simon E. Fisher, Philippe M. Campeau. A full list of consortium members appears below.

The DDD study

Jeremy F. McRae⁴⁸, Stephen Clayton⁴⁸, Tomas W. Fitzgerald⁴⁸, Joanna Kaplanis⁴⁸, Elena Prigmore⁴⁸, Diana Rajan⁴⁸, Alejandro Sifrim⁴⁸, Stuart Aitken⁴⁹, Nadia Akawi⁴⁸, Mohsan Alvi⁵⁰, Kirsty Ambridge⁴⁸, Daniel M. Barrett⁴⁸, Tanya Bayzetinova⁴⁸, Philip Jones⁴⁸, Wendy D. Jones⁴⁸, Daniel King⁴⁸, Netravathi Krishnappa⁴⁸, Laura E. Mason⁴⁸, Tarjinder Singh⁴⁸, Adrian R. Tivey⁴⁸, Munaza Ahmed^{51,52,53}, Uruj Anjum⁵⁴, Hayley Archer^{55,56}, Ruth Armstrong⁵⁷, Jana Awada⁴⁸, Meena Balasubramanian⁵⁸, Siddharth Banka⁵⁹, Diana Baralle^{51,52,53}, Angela Barnicoat⁶⁰, Paul Batstone⁶¹, David Baty⁶², Chris Bennett⁶³, Jonathan Berg⁶², Birgitta Bernhard⁶⁴, A. Paul Bevan⁴⁸, Maria Bitner-Glindzicz⁶⁰, Edward Blair⁶⁵, Moira Blyth⁶³, David Bohanna⁶⁶, Louise Bourdon⁶⁴, David Bourn⁶⁷, Lisa Bradley⁶⁸, Angela Brady⁶⁴, Simon Brent⁴⁸, Carole Brewer⁶⁹, Kate Brunstrom⁶⁰, David J. Bunyan^{51,52,53}, John Burn⁶⁷, Natalie Canham⁶⁴, Bruce Castle⁶⁹, Kate Chandler⁵⁹, Elena Chatzimichali⁴⁸, Deirdre Cilliers⁶⁵, Angus Clarke^{55,56}, Susan Clasper⁶⁵, Jill Clayton-Smith⁵⁹, Virginia Clowes⁶⁴, Andrea Coates⁶³, Trevor Cole⁶⁶, Irina Colgiu⁴⁸, Amanda Collins^{51,52,53},

Morag N. Collinson^{51,52,53}, Fiona Connell⁷⁰, Nicola Cooper⁶⁶, Helen Cox⁶⁶, Lara Cresswell⁷¹, Gareth Cross⁷², Yanick Crow⁵⁹, Mariella D'Alessandro⁶¹, Tabib Dabir⁶⁸, Rosemarie Davidson⁷³, Sally Davies^{55,56}, Dylan de Vries⁴⁸, John Dean⁶¹, Charu Deshpande⁷⁰, Gemma Devlin⁶⁹, Abhijit Dixit⁷², Angus Dobbie⁶³, Alan Donaldson⁷⁴, Dian Donnai⁵⁹, Deirdre Donnelly⁶⁸, Carina Donnelly⁵⁹, Angela Douglas⁷⁵, Sofia Douzou⁵⁹, Alexis Duncan⁷³, Jacqueline Eason⁷², Sian Ellard⁶⁹, Ian Ellis⁷⁵, Frances Elmslie⁵⁴, Karenza Evans^{55,56}, Sarah Everest⁶⁹, Tina Fendick⁷⁰, Richard Fisher⁶⁷, Frances Flinter⁷⁰, Nicola Foulds^{51,52,53}, Andrew Fry^{55,56}, Alan Fryer⁷⁵, Carol Gardiner⁷³, Lorraine Gaunt⁵⁹, Neeti Ghali⁶⁴, Richard Gibbons⁶⁵, Harinder Gill⁷⁶, Judith Goodship⁶⁷, David Goudie⁶², Emma Gray⁴⁸, Andrew Green⁷⁶, Philip Greene⁴⁸, Lynn Greenhalgh⁷⁵, Susan Gribble⁴⁸, Rachel Harrison⁷², Lucy Harrison^{51,52,53}, Victoria Harrison^{51,52,53}, Rose Hawkins⁷⁴, Liu He⁴⁸, Stephen Hellens⁶⁷, Alex Henderson⁶⁷, Sarah Hewitt⁶³, Lucy Hildyard⁴⁸, Emma Hobson⁶³, Simon Holden⁵⁷, Muriel Holder⁶⁴, Susan Holder⁶⁴, Georgina Hollingsworth⁶⁰, Tessa Homfray⁵⁴, Mervyn Humphreys⁶⁸, Jane Hurst⁶⁰, Ben Hutton⁴⁸, Stuart Ingram⁵⁸, Melita Irving⁷⁰, Lily Islam⁶⁶, Andrew Jackson⁴⁹, Joanna Jarvis⁶⁶, Lucy Jenkins⁶⁰, Diana Johnson⁵⁸, Elizabeth Jones⁵⁹, Dragana Josifova⁷⁰, Shelagh Joss⁷³, Beckie Kaemba⁷¹, Sandra Kazembe⁷¹, Rosemary Kelsell⁴⁸, Bronwyn Kerr⁵⁹, Helen Kingston⁵⁹, Usha Kini⁶⁵, Esther Kinning⁷³, Gail Kirby⁶⁶, Claire Kirk⁶⁸, Emma Kivuva⁶⁹, Alison Kraus⁶³, Dhavendra Kumar^{55,56}, V.K.Ajith Kumar⁶⁰, Katherine Lachlan^{51,52,53}, Wayne Lam⁴⁹, Anne Lampe⁴⁹, Caroline Langman⁷⁰, Melissa Lees⁶⁰, Derek Lim⁶⁶, Cheryl Longman⁷³, Gordon Lowther⁷³, Sally A. Lynch⁷⁶, Alex Magee⁶⁸, Eddy Maher⁴⁹, Alison Male⁶⁰, Sahar Mansour⁵⁴, Karen Marks⁵⁴, Katherine Martin⁷², Una Maye⁷⁵, Emma McCann⁷⁷, Vivienne McConnell⁶⁸, Meriel McEntagart⁵⁴, Ruth McGowan⁶¹, Kirsten McKay⁶⁶, Shane McKee⁶⁸, Dominic J. McMullan⁶⁶, Susan McNerlan⁶⁸, Catherine McWilliam⁶¹, Sarju Mehta⁵⁷, Kay Metcalfe⁵⁹, Anna Middleton⁴⁸, Zosia Miedzymbrodzka⁶¹, Emma Miles⁵⁹, Shehla Mohammed⁷⁰, Tara Montgomery⁶⁷, David Moore⁴⁹, Sian Morgan^{55,56}, Jenny Morton⁶⁶, Hood Mugalaasi^{55,56}, Victoria Murday⁷³, Helen Murphy⁵⁹, Swati Naik⁶⁶, Andrea Nemeth⁶⁵, Louise Nevitt⁵⁸, Andrew Norman⁶⁶, Rosie O'Shea⁷⁶, Caroline Ogilvie⁷⁰, Kai-Ren Ong⁶⁶, Soo-Mi Park⁵⁷, Michael J. Parker⁵⁸, Chirag Patel⁶⁶, Joan Paterson⁵⁷, Stewart Payne⁶⁴, Daniel Perrett⁴⁸, Julie Phipps⁶⁵, Daniela T. Pilz⁷³, Martin Pollard⁴⁸, Caroline Pottinger⁷⁷, Joanna Poulton⁶⁵, Norman Pratt⁶², Katrina Prescott⁶³, Abigail Pridham⁶⁵, Annie Procter^{55,56}, Hellen Purnell⁶⁵, Oliver Quarrell⁵⁸, Nicola Ragge⁶⁶, Raheleh Rahbari⁴⁸, Josh Randall⁴⁸, Lucy Raymond⁵⁷, Debbie Rice⁶², Leema Robert⁷⁰, Eileen Roberts⁷⁴, Jonathan Roberts⁵⁷, Paul Roberts⁶³, Gillian Roberts⁷⁵, Alison Ross⁶¹, Elisabeth Rosser⁶⁰, Anand Saggar⁵⁴, Shalaka Samant⁶¹, Julian Sampson^{55,56}, Richard Sandford⁵⁷, Ajoy Sarkar⁷², Susann Schweiger⁶², Richard Scott⁶⁰, Ingrid Scurr⁷⁴, Ann Selby⁷², Anneke Seller⁶⁵, Cheryl Sequeira⁶⁴, Nora Shannon⁷², Saba Sharif⁶⁶, Charles Shaw-Smith⁶⁹, Emma Shearing⁵⁸, Debbie Shears⁶⁵, Eamonn Sheridan⁶³, Ingrid Simonic⁵⁷, Roldan Singzon⁶⁴, Zara Skitt⁵⁹, Audrey Smith⁶³, Kath Smith⁵⁸, Sarah Smithson⁷⁴, Linda Sneddon⁶⁷, Miranda Splitt⁶⁷, Miranda Squires⁶³, Fiona Stewart⁶⁸, Helen Stewart⁶⁵, Volker Straub⁶⁷, Mohnish Suri⁷², Vivienne Sutton⁷⁵, Ganesh Jawahar Swaminathan⁴⁸, Elizabeth Sweeney⁷⁵, Kate Tatton-Brown⁵⁴, Cat Taylor⁵, Rohan Taylor⁵⁴, Mark Tein⁶⁶, I. Karen Temple^{51,52,53}, Jenny Thomson⁶³, Marc Tischkowitz⁵⁷, Susan Tomkins⁷⁴, Audrey Torokwa^{51,52,53}, Becky Treacy⁵⁷, Claire Turner⁶⁹, Peter Turnpenny⁶⁹, Carolyn Tysoe⁶⁹, Anthony Vandersteen⁶⁴, Vinod Varghese^{55,56}, Pradeep Vasudevan⁷¹, Parthiban Vijayarangakannan⁴⁸, Julie Vogt⁶⁶, Emma Wakeling⁶⁴, Sarah Wallwark⁵⁷, Jonathon Waters⁶⁰, Astrid Weber⁷⁵, Diana Wellesley^{51,52,53}, Margo Whiteford⁷³, Sara Widaa⁴⁸, Sarah Wilcox⁵⁷, Emily Wilkinson⁴⁸, Denise Williams⁶⁶, Nicola Williams⁷³, Louise Wilson⁶⁰, Geoff Woods⁵⁷, Christopher Wragg⁷⁴, Michael Wright⁶⁷, Laura Yates⁶⁷, Michael Yau⁷⁰, Chris Nellåker^{78,79,80}, Michael Parker⁸¹, Helen V. Firth^{48,57}, Caroline F. Wright⁴⁸, David R. FitzPatrick^{48,49}, Jeffrey C. Barrett⁴⁸ & Matthew E. Hurles⁴⁸

⁴⁸Wellcome Trust Sanger Institute, Wellcome Trust Genome Campus, Hinxton, Cambridge CB10 1SA, UK. ⁴⁹MRC Human Genetics Unit, MRC IGMM, University of Edinburgh, Western General Hospital, Edinburgh EH4 2XU, UK. ⁵⁰Department of Engineering Science, University of Oxford, Parks Road, Oxford OX1 3PJ, UK. ⁵¹Wessex Clinical Genetics Service, University Hospital Southampton, Princess Anne Hospital, Coxford Road, Southampton SO16 5YA, UK. ⁵²Wessex Regional Genetics Laboratory, Salisbury NHS Foundation Trust, Salisbury District Hospital, Odstock Road, Salisbury, Wiltshire SP2 8BJ, UK. ⁵³Faculty of Medicine, University of Southampton, Building 85, Life Sciences Building, Highfield Campus, Southampton SO17 1BJ, UK. ⁵⁴South West Thames Regional Genetics Centre, St George's Healthcare NHS Trust, St George's, University of London, Cranmer Terrace, London SW17 0RE, UK. ⁵⁵Institute of Medical Genetics, University Hospital of Wales, Heath Park, Cardiff CF14 4XW, UK. ⁵⁶Department of Clinical Genetics, Block 12, Glan Clwyd Hospital, Rhyl, Denbighshire LL18 5UJ, UK. ⁵⁷East Anglian Medical Genetics Service, Box 134, Cambridge University Hospitals NHS Foundation Trust, Cambridge Biomedical Campus, Cambridge CB2 0QQ, UK. ⁵⁸Sheffield Regional Genetics Services, Sheffield Children's NHS Trust, Western Bank, Sheffield S10 2TH, UK. ⁵⁹Manchester Centre for Genomic Medicine, St Mary's Hospital, Central Manchester University Hospitals NHS Foundation Trust, Manchester Academic Health Science Centre, Manchester M13 9WL, UK. ⁶⁰North East Thames Regional Genetics Service, Great Ormond Street Hospital for Children NHS Foundation Trust, Great Ormond Street Hospital, Great Ormond Street, London WC1N3JH, UK. ⁶¹North of Scotland Regional Genetics Service, NHS Grampian, Department of Medical Genetics Medical School, Foresterhill, Aberdeen AB25 2ZD, UK. ⁶²East of Scotland Regional Genetics Service, Human Genetics Unit, Pathology Department, NHS Tayside, Ninewells Hospital, Dundee DD1 9SY, UK. ⁶³Yorkshire Regional Genetics Service, Leeds Teaching Hospitals NHS Trust, Department of Clinical Genetics, Chapel Allerton Hospital, Chapeltown Road, Leeds LS7 4SA, UK. ⁶⁴North West Thames Regional Genetics Centre, North West London Hospitals NHS Trust, The Kennedy Galton Centre, Northwick Park and St Mark's NHS Trust Watford Road, Harrow HA1 3UJ, UK. ⁶⁵Oxford Regional Genetics Service, Oxford Radcliffe Hospitals NHS Trust, The Churchill Old Road, Oxford OX3 7LJ, UK. ⁶⁶West Midlands Regional Genetics Service, Birmingham Women's NHS Foundation Trust, Birmingham Women's Hospital, Edgbaston, Birmingham B15 2TG, UK. ⁶⁷Northern Genetics Service, Newcastle upon Tyne Hospitals NHS Foundation Trust, Institute of Human Genetics, International Centre for Life, Central Parkway, Newcastle upon Tyne NE1 3BZ, UK. ⁶⁸Northern Ireland Regional Genetics Centre, Belfast Health and Social Care Trust, Belfast City Hospital, Lisburn Road, Belfast BT9 7AB, UK. ⁶⁹Peninsula Clinical Genetics Service, Royal Devon and Exeter NHS Foundation Trust, Clinical Genetics Department, Royal Devon & Exeter Hospital (Heavitree), Gladstone Road, Exeter EX1 2ED, UK. ⁷⁰South East Thames Regional Genetics Centre, Guy's and St Thomas' NHS Foundation Trust, Guy's Hospital, Great Maze Pond, London SE1 9RT, UK. ⁷¹Leicestershire Genetics Centre, University Hospitals of Leicester NHS Trust, Leicester Royal Infirmary (NHS Trust), Leicester LE1 5WW, UK. ⁷²Nottingham Regional Genetics Service, City Hospital Campus, Nottingham University Hospitals NHS Trust, The Gables, Hucknall Road, Nottingham NG5 1PB, UK. ⁷³West of Scotland Regional Genetics Service, NHS Greater Glasgow and Clyde, Institute of Medical Genetics, Yorkhill Hospital, Glasgow G3 8SJ, UK. ⁷⁴Bristol Genetics Service (Avon, Somerset, Gloucs and West Wilts), University Hospitals Bristol NHS Foundation Trust, St Michael's Hospital, St Michael's Hill, Bristol BS2 8DT, UK. ⁷⁵Merseyside and Cheshire Genetics Service, Liverpool Women's NHS Foundation Trust, Department of Clinical Genetics, Royal Liverpool Children's Hospital Alder Hey, Eaton Road, Liverpool L12 2AP, UK. ⁷⁶National Centre for Medical Genetics, Our Lady's Children's Hospital, Crumlin, Dublin 12, Ireland. ⁷⁷Department of Clinical Genetics, Block 12, Glan Clwyd Hospital, Rhyl, Denbighshire LL18 5UJ Wales, UK. ⁷⁸Nuffield Department of Obstetrics & Gynaecology, University of Oxford, Level 3, Women's Centre, John Radcliffe Hospital, Oxford OX3 9DU, UK. ⁷⁹Institute of Biomedical Engineering, Department of Engineering Science, University of Oxford, Old Road Campus Research Building, Oxford OX3 7DQ, UK. ⁸⁰Big Data Institute, University of Oxford, Roosevelt drive, Oxford OX3 7LF, UK. ⁸¹The Ethox Centre, Nuffield Department of Population Health, University of Oxford, Old Road Campus, Oxford OX3 7LF, UK