



This is a repository copy of *Genomic sequencing technologies for rare disease in mainstream healthcare: the current state of implementation*.

White Rose Research Online URL for this paper:

<https://eprints.whiterose.ac.uk/id/eprint/231820/>

Version: Supplemental Material

Article:

Mackley, M.P. orcid.org/0000-0002-7388-0905, Agrawal, P.B. orcid.org/0000-0003-3255-0456, Ali, S.S. et al. (15 more authors) (2025) Genomic sequencing technologies for rare disease in mainstream healthcare: the current state of implementation. *European Journal of Human Genetics*. pp. 1-12. ISSN: 1018-4813

<https://doi.org/10.1038/s41431-025-01925-7>

© 2025 The Authors. Except as otherwise noted, this author-accepted version of a journal article published in *European Journal of Human Genetics* is made available via the University of Sheffield Research Publications and Copyright Policy under the terms of the Creative Commons Attribution 4.0 International License (CC-BY 4.0), which permits unrestricted use, distribution and reproduction in any medium, provided the original work is properly cited. To view a copy of this licence, visit <http://creativecommons.org/licenses/by/4.0/>

Reuse

This article is distributed under the terms of the Creative Commons Attribution (CC BY) licence. This licence allows you to distribute, remix, tweak, and build upon the work, even commercially, as long as you credit the authors for the original work. More information and the full terms of the licence here: <https://creativecommons.org/licenses/>

Takedown

If you consider content in White Rose Research Online to be in breach of UK law, please notify us by emailing eprints@whiterose.ac.uk including the URL of the record and the reason for the withdrawal request.



eprints@whiterose.ac.uk
<https://eprints.whiterose.ac.uk/>

Supplement 1. Additional references

Below are additional references sorted by section:

ROUTINE DIAGNOSTIC GENOMIC TESTING

Implementation of genomic testing in general paediatrics

1. Ashtiani S, Makela N, Carrion P, Austin J. Parents' experiences of receiving their child's genetic diagnosis: a qualitative study to inform clinical genetics practice. *Am J Med Genet A*. 2014;164A(6):1496-502.
2. Griffin BH, Chitty LS, Bitner-Glindzicz M. The 100 000 Genomes Project: What it means for paediatrics. *Arch Dis Child Educ Pract Ed*. 2017;102(2):105-7.
3. Klitzman R, Bezborodko E, Chung WK, Appelbaum PS. Parents' views of benefits and limitations of receiving genetic diagnoses for their offspring. *Child Care Health Dev*. 2024;50(1):e13212.
4. Lewis C, Sanderson S, Hill M, Patch C, Searle B, Hunter A, et al. Parents' motivations, concerns and understanding of genome sequencing: a qualitative interview study. *Eur J Hum Genet*. 2020;28(7):874-84.
5. Manickam K, McClain MR, Demmer LA, Biswas S, Kearney HM, Malinowski J, et al. Exome and genome sequencing for pediatric patients with congenital anomalies or intellectual disability: an evidence-based clinical guideline of the American College of Medical Genetics and Genomics (ACMG). *Genet Med*. 2021;23(11):2029-37.
6. Peter M, Hammond J, Sanderson SC, Gurasashvili J, Hunter A, Searle B, et al. Participant experiences of genome sequencing for rare diseases in the 100,000 Genomes Project: a mixed methods study. *Eur J Hum Genet*. 2022;30(5):604-10.
7. Frankish N. Good diagnosis: improving the experiences of diagnosis for people with rare conditions. . London: Rare Disease UK. 2022.
8. NHS England. National Genomic Test Directory for rare and inherited disease. In: England N, editor. 2022.
9. Medical Services Advisory Committee. 1476-Genetic testing for childhood syndromes. 2021.
10. Ellard H, Clarke A, Wynn S, Pichini A, Lewis C. Written communication of whole genome sequencing results in the NHS Genomic Medicine Service: a multi-centre service evaluation. *Eur J Hum Genet*. 2024;32(11):1436-45.

Implementation of genomic testing in nephrology

11. Elhassan EAE, Murray SL, Connaughton DM, Kennedy C, Cormican S, Cowhig C, et al. The utility of a genetic kidney disease clinic employing a broad range of genomic testing platforms: experience of the Irish Kidney Gene Project. *J Nephrol*. 2022;35(6):1655-65.
12. Connaughton DM, Hildebrandt F. Personalized medicine in chronic kidney disease by detection of monogenic mutations. *Nephrol Dial Transplant*. 2020;35(3):390-7.

13. Groopman EE, Marasa M, Cameron-Christie S, et al. Diagnostic Utility of Exome Sequencing for Kidney Disease. *The New England journal of medicine*. 2019.
14. Jayasinghe K, Biros E, Harris T, Wood A, O'Shea R, Hill L, et al. Implementation and Evaluation of a National Multidisciplinary Kidney Genetics Clinic Network Over 10 Years. *Kidney Int Rep*. 2024;9(8):2372-85.
15. Jayasinghe K, Stark Z, Kerr PG, Gaff C, Martyn M, Whitlam J, et al. Clinical impact of genomic testing in patients with suspected monogenic kidney disease. *Genet Med*. 2021;23(1):183-91.
16. Chen Y, Zhang Y, Huang J, Zeng Y, Qian Y, Chen J, et al. New insights from trio whole-exome sequencing in the children with kidney disease: A single-center retrospective cohort study. *Mol Genet Genomic Med*. 2023;11(7):e2163.
17. Schott C, Lebedeva V, Taylor C, Abumelha S, Roshanov PS, Connaughton DM. Utility of Genetic Testing in Adults with CKD: A Systematic Review and Meta-Analysis. *Clin J Am Soc Nephrol*. 2025;20(1):101-15.
18. Schott C, Alajmi M, Bukhari M, Relouw S, Wang J, McIntyre AD, et al. Genetic Testing in Adults over 50 Years with Chronic Kidney Disease: Diagnostic Yield and Clinical Implications in a Specialized Kidney Genetics Clinic. *Genes (Basel)*. 2025;16(4).
19. Wu Y, Jayasinghe K, Stark Z, Quinlan C, Patel C, McCarthy H, et al. Genomic testing for suspected monogenic kidney disease in children and adults: A health economic evaluation. *Genet Med*. 2023;25(11):100942.
20. Jayasinghe K, Wu Y, Stark Z, Kerr PG, Mallett AJ, Gaff C, et al. Cost-Effectiveness of Targeted Exome Analysis as a Diagnostic Test in Glomerular Diseases. *Kidney Int Rep*. 2021;6(11):2850-61.
21. Health] AGDo. Medicare Benefits Schedule Book Operating from 1 March 2020. 2020.
22. KDIGO Clinical Practice Guideline for the Evaluation, Management, and Treatment of Autosomal Dominant Polycystic Kidney Disease. *Kidney Int*. 2025.
23. Floege J, Gibson KL, Vivarelli M, Liew A, Radhakrishnan J, Balk EM, et al. Executive summary of the KDIGO 2025 Clinical Practice Guideline for the Management of Nephrotic Syndrome in Children. *Kidney Int*. 2025;107(5):806-8.

RAPID DIAGNOSTIC GENOMIC TESTING IN INTENSIVE CARE

24. Wojcik MH, Lemire G, Berger E, Zaki MS, Wissmann M, Win W, et al. Genome Sequencing for Diagnosing Rare Diseases. *N Engl J Med*. 2024;390(21):1985-97.
25. Marom D, Mory A, Reytan-Miron S, Amir Y, Kurolap A, Cohen JG, et al. National Rapid Genome Sequencing in Neonatal Intensive Care. *JAMA Netw Open*. 2024;7(2):e240146.
26. van Diemen CC, Kerstjens-Frederikse WS, Bergman KA, de Koning TJ, Sikkema-Raddatz B, van der Velde JK, et al. Rapid Targeted Genomics in Critically Ill Newborns. *Pediatrics*. 2017;140(4).
27. Petrikin JE, Cakici JA, Clark MM, Willig LK, Sweeney NM, Farrow EG, et al. The NSIGHT1-randomized controlled trial: rapid whole-genome sequencing for accelerated etiologic diagnosis in critically ill infants. *NPJ Genom Med*. 2018;3:6.

28. Farnaes L, Hildreth A, Sweeney NM, Clark MM, Chowdhury S, Nahas S, et al. Rapid whole-genome sequencing decreases infant morbidity and cost of hospitalization. *NPJ Genom Med.* 2018;3:10.
29. Dimmock DP, Clark MM, Gaughran M, Cakici JA, Caylor SA, Clarke C, et al. An RCT of Rapid Genomic Sequencing among Seriously Ill Infants Results in High Clinical Utility, Changes in Management, and Low Perceived Harm. *Am J Hum Genet.* 2020.
30. Dimmock D, Caylor S, Waldman B, Benson W, Ashburner C, Carmichael JL, et al. Project Baby Bear: Rapid precision care incorporating rWGS in 5 California children's hospitals demonstrates improved clinical outcomes and reduced costs of care. *Am J Hum Genet.* 2021;108(7):1231-8.
31. Group TNS. Effect of Whole-Genome Sequencing on the Clinical Management of Acutely Ill Infants With Suspected Genetic Disease: A Randomized Clinical Trial. *JAMA Pediatrics.* 2021.
32. Denomme-Pichon AS, Vitobello A, Olaso R, Ziegler A, Jeanne M, Tran Mau-Them F, et al. Accelerated genome sequencing with controlled costs for infants in intensive care units: a feasibility study in a French hospital network. *Eur J Hum Genet.* 2022;30(5):567-76.
33. Mestek-Boukhibar L, Clement E, Jones WD, Drury S, Ocaka L, Gagunashvili A, et al. Rapid Paediatric Sequencing (RaPS): comprehensive real-life workflow for rapid diagnosis of critically ill children. *J Med Genet.* 2018;55(11):721-8.
34. French CE, Delon I, Dolling H, Sanchis-Juan A, Shamardina O, Mégy K, et al. Whole genome sequencing reveals that genetic conditions are frequent in intensively ill children. *Intensive Care Med.* 2019.
35. Wang H, Lu Y, Dong X, Lu G, Cheng G, Qian Y, et al. Optimized trio genome sequencing (OTGS) as a first-tier genetic test in critically ill infants: practice in China. *Hum Genet.* 2020.
36. Diaby V, Babcock A, Huang Y, Moussa RK, Espinal PS, Janvier M, et al. Real-world economic evaluation of prospective rapid whole-genome sequencing compared to a matched retrospective cohort of critically ill pediatric patients in the United States. *Pharmacogenomics J.* 2022;22(4):223-9.
37. Lumaka A, Fasquelle C, Debray FG, Alkan S, Jacquinet A, Harvengt J, et al. Rapid Whole Genome Sequencing Diagnoses and Guides Treatment in Critically Ill Children in Belgium in Less than 40 Hours. *Int J Mol Sci.* 2023.
38. Brett GR, Martyn M, Lynch F, de Silva MG, Ayres S, Gallacher L, et al. Parental experiences of ultrarapid genomic testing for their critically unwell infants and children. *Genet Med.* 2020;22(12):1976-85.
39. Cakici JA, Dimmock DP, Caylor SA, Gaughran M, Clarke C, Triplett C, et al. A Prospective Study of Parental Perceptions of Rapid Whole-Genome and -Exome Sequencing among Seriously Ill Infants. *Am J Hum Genet.* 2020;107(5):953-62.
40. Lynch F, Nisselle A, Gaff CL, McClaren B. Rapid acute care genomics: Challenges and opportunities for genetic counselors. *J Genet Couns.* 2021;30(1):30-41.
41. Lynch F, Nisselle A, Stark Z, Gaff CL, McClaren B. Parents' experiences of decision making for rapid genomic sequencing in intensive care. *Eur J Hum Genet.* 2021;29(12):1804-10.

42. Smith EE, du Souich C, Dragojlovic N, Study C, Study R, Elliott AM. Genetic counseling considerations with rapid genome-wide sequencing in a neonatal intensive care unit. *J Genet Couns*. 2019;28(2):263-72.
43. Berrios C, Koertje C, Noel-MacDonnell J, Soden S, Lantos J. Parents of newborns in the NICU enrolled in genome sequencing research: hopeful, but not naive. *Genet Med*. 2020;22(2):416-22.
44. Lemke AA, Thompson ML, Gimpel EC, McNamara KC, Rich CA, Finnila CR, et al. Parents' Perspectives on the Utility of Genomic Sequencing in the Neonatal Intensive Care Unit. *Journal of personalized medicine*. 2023;13(7).
45. Callahan KP, Clayton EW, Lemke AA, Chaudhari BP, Wenger TL, Lyle ANJ, et al. Ethical and Legal Issues Surrounding Genetic Testing in the NICU. *Neoreviews*. 2024;25(3):e127-e38.
46. Gal DB, Deutch N, Lee SSJ, Simon RT, Char DS. Parental Attitudes Toward Clinical Genomic Sequencing in Children With Critical Cardiac Disease. *Pediatr Crit Care Med*. 2021;22(8):e419-e26.
47. Callahan KP, Mueller R, Joffe S, Skraban C, Spinner N, Crew K, et al. Parents' Perceptions of the Utility of Genetic Testing in the NICU. *Genet Med*. 2025:101393.
48. Bowman-Smart H, Vears DF, Brett GR, Martyn M, Stark Z, Gyngell C. 'Diagnostic shock': the impact of results from ultrarapid genomic sequencing of critically unwell children on aspects of family functioning. *Eur J Hum Genet*. 2022;30(9):1036-43.
49. Clowes Candadai SV, Sikes MC, Thies JM, Freed AS, Bennett JT. Rapid clinical exome sequencing in a pediatric ICU: Genetic counselor impacts and challenges. *J Genet Couns*. 2019;28(2):283-91.
50. Boggs K, Lynch F, Ward M, Bouffler SE, Ayres S, Forbes R, et al. Rapid genomic testing in critically ill pediatric patients: Genetic counseling lessons from a national program. *Genet Med Open*. 2024;2(Suppl 2):101878.
51. Kingsmore SF, Cole FS. The Role of Genome Sequencing in Neonatal Intensive Care Units. *Annu Rev Genomics Hum Genet*. 2022;23:427-48.
52. Saunders CJ, Miller NA, Soden SE, Dinwiddie DL, Noll A, Alnadi NA, et al. Rapid whole-genome sequencing for genetic disease diagnosis in neonatal intensive care units. *Sci Transl Med*. 2012;4(154):154ra35.
53. Goenka SD, Gorzynski JE, Shafin K, Fisk DG, Pesout T, Jensen TD, et al. Accelerated identification of disease-causing variants with ultra-rapid nanopore genome sequencing. *Nat Biotechnol*. 2022;40(7):1035-41.
54. Miller NA, Farrow EG, Gibson M, Willig LK, Twist G, Yoo B, et al. A 26-hour system of highly sensitive whole genome sequencing for emergency management of genetic diseases. *Genome Med*. 2015;7:100.

GENOMIC NEWBORN SCREENING

55. Ceyhan-Birsoy O, Murry JB, Machini K, Lebo MS, Yu TW, Fayer S, et al. Interpretation of Genomic Sequencing Results in Healthy and Ill Newborns: Results from the BabySeq Project. *The American Journal of Human Genetics*. 2019;104(1):76-93.

56. Smith HS, Zettler B, Genetti CA, Hickingbotham MR, Coleman TF, Lebo M, et al. The BabySeq Project: A clinical trial of genome sequencing in a diverse cohort of infants. *The American Journal of Human Genetics*. 2024;111(10):2094-106.
57. Adhikari AN, Gallagher RC, Wang Y, Currier RJ, Amatuni G, Bassaganyas L, et al. The role of exome sequencing in newborn screening for inborn errors of metabolism. *Nature Medicine*. 2020;26(9):1392-7.
58. Roman TS, Crowley SB, Roche MI, Foreman AKM, O'Daniel JM, Seifert BA, et al. Genomic Sequencing for Newborn Screening: Results of the NC NEXUS Project. *The American Journal of Human Genetics*. 2020;107(4):596-611.
59. Kingsmore SF, Smith LD, Kunard CM, Bainbridge M, Batalov S, Benson W, et al. A genome sequencing system for universal newborn screening, diagnosis, and precision medicine for severe genetic diseases. *The American Journal of Human Genetics*. 2022;109(9):1605-19.
60. Ziegler A, Koval-Burt C, Kay DM, Suchy SF, Begtrup A, Langley KG, et al. Expanded Newborn Screening Using Genome Sequencing for Early Actionable Conditions. *JAMA*. 2025;333(1).
61. Sullivan J, Jalazo E, Powell C, Cope H, Forsythe A, Kucera K, et al. P590: Unique outcomes from curated genome sequencing for newborn screening: Challenging results from the Early Check project. *Genet Med Open*. 2025;3:102438.
62. Cope HL, Milko LV, Jalazo ER, Crissman BG, Foreman AKM, Powell BC, et al. A systematic framework for selecting gene-condition pairs for inclusion in newborn sequencing panels: Early Check implementation. *Genetics in Medicine*. 2024;26(12):101290.
63. Redit C, Hura Z, Amy L. UK launches whole-genome sequencing pilot for babies. *Nat Biotechnol*. 2023 Jan;41:3-8.
64. Garnier N, Berghout J, Zygmunt A, Singh D, Huang KA, Kantz W, et al. Genetic newborn screening and digital technologies: A project protocol based on a dual approach to shorten the rare diseases diagnostic path in Europe. *PLOS ONE*. 2023;18(11):e0293503.
65. Ferlini A, Gross ES, Garnier N, Berghout J, Zygmunt A, Singh D, et al. Rare diseases' genetic newborn screening as the gateway to future genomic medicine: the Screen4Care EU-IMI project. *Orphanet Journal of Rare Diseases*. 2023;18(1):310.
66. Boemer F, Hovhannesian K, Piazzon F, Minner F, Mni M, Jacquemin V, et al. Population-based, first-tier genomic newborn screening in the maternity ward. *Nature Medicine*. 2025:1-12.
67. Ji C, Farrar MA, Norris S, Bhattacharya K, Bennetts B, Newson AJ, et al. The Australian landscape of newborn screening in the genomics era. *Rare Dis Orphan Drugs J*. 2023;2(4):null-null.
68. Lunke S, Bouffler SE, Downie L, Caruana J, Amor DJ, Archibald A, et al. Prospective cohort study of genomic newborn screening: BabyScreen+ pilot study protocol. *BMJ Open*. 2024;14(4):e081426.
69. Kassahn KS, Anastasi LT, Chowdhury A, Ashenden A, Skinner S, Rozek T, et al. NewbornsInSA: multi-omic model for genomic newborn screening. *Pathology*. 2025;57:S17-S8.

70. Hao C, Guo R, Hu X, Qi Z, Guo Q, Liu X, et al. Newborn screening with targeted sequencing: a multicenter investigation and a pilot clinical study in China. *J Genet Genom.* 2022;49(1):13-9.
71. Chen T, Fan C, Huang Y, Feng J, Zhang Y, Miao J, et al. Genomic Sequencing as a First-Tier Screening Test and Outcomes of Newborn Screening. *JAMA Netw Open.* 2023;6(9):e2331162.
72. Kelly NR, Orsini JJ, Goldenberg AJ, Mulrooney NS, Boychuk NA, Clarke MJ, et al. ScreenPlus: A comprehensive, multi-disorder newborn screening program. *Mol Genet Metab Rep.* 2024;38:101037.

GENETIC COUNSELLING OUTSIDE THE CLINICAL GENETICS SERVICE

73. Kirk EP, Delatycki MB, Archibald AD, Tutty E, Caruana J, Halliday JL, et al. Nationwide, Couple-Based Genetic Carrier Screening. *N Engl J Med.* 2024;391(20):1877-89.
74. Birnie E, Schuurmans J, Plantinga M, Abbott KM, Fenwick A, Lucassen A, et al. Couple-based expanded carrier screening provided by general practitioners to couples in the Dutch general population: psychological outcomes and reproductive intentions. *Genet Med.* 2021;23(9):1761-8.
75. Kraft SA, Schneider JL, Leo MC, Kauffman TL, Davis JV, Porter KM, et al. Patient actions and reactions after receiving negative results from expanded carrier screening. *Clin Genet.* 2018;93(5):962-71.
76. Van Steijvoort E, Peeters H, Vandecruys H, Verguts J, Peeraer K, Matthijs G, et al. Experiences of nonpregnant couples after receiving reproductive genetic carrier screening results in Belgium. *Eur J Hum Genet.* 2023;31(6):696-702.
77. Beard CA, Amor DJ, Di Pietro L, Archibald AD. "I'm Healthy, It's Not Going To Be Me": Exploring experiences of carriers identified through a population reproductive genetic carrier screening panel in Australia. *Am J Med Genet.* 2016.
78. Ioannou L, Delatycki MB, Massie J, Hodgson J, Lewis S. "Suddenly Having two Positive People who are Carriers is a Whole New Thing" - Experiences of Couples Both Identified as Carriers of Cystic Fibrosis Through a Population-Based Carrier Screening Program in Australia. *J Genet Couns.* 2015.
79. Richardson E, McEwen A, Newton-John T, Crook A, Jacobs C. Outcomes of Importance to Patients in Reproductive Genetic Carrier Screening: A Qualitative Study to Inform a Core Outcome Set. *Journal of personalized medicine.* 2022;12(8).
80. Rothwell E, Johnson E, Mathiesen A. Experiences among Women with Positive Prenatal Expanded Carrier Screening Results. *Journal of genetic counseling.* 2017.
81. Best S, Long JC, Fehlberg Z. The more you do it, the easier it gets: using behaviour change theory to support health care professionals offering reproductive genetic carrier screening. *Eur J Hum Genet.* 2023.
82. Alexander KE, Rolfe M, Gabbett MT. Assessing genomics confidence and learning needs in Australian nurses and midwives: an educational program evaluation. *Front Genet.* 2024;15:1419302.
83. Gusen T, Freeman L, Musgrave L. Empowering midwives with genetic knowledge: A systematic review of educational needs in genomics. *Nurse Educ Pract.* 2025;84:104340.

84. Lee YQ, Yoon SY, Hassan T, Padmanabhan H, Yip CH, Keng WT, et al. Attitudes and training needs of oncologists and surgeons in mainstreaming breast cancer genetic counseling in a low-to-middle income Asian country. *J Genet Couns*. 2022;31(5):1080-9.
85. Nisselle A, King EA, McClaren B, Janinski M, Metcalfe S, Gaff C, et al. Measuring physician practice, preparedness and preferences for genomic medicine: a national survey. *BMJ Open*. 2021;11(7):e044408.
86. Seed L, Scott A, Peter M, Tadros S, Hill M, da Costa CS. Preparing tomorrow's doctors for the genomics era: A nationwide survey of UK medical students. *Future Healthc J*. 2024;11(2):100133.
87. Yanes T, Sullivan A, Barbaro P, Brion K, Hollway G, Peake J, et al. Evaluation and pilot testing of a multidisciplinary model of care to mainstream genomic testing for paediatric inborn errors of immunity. *Eur J Hum Genet*. 2023.

WORKFORCE DEVELOPMENT CONSIDERATIONS FOR MAINSTREAM GENOMIC MEDICINE

Genomics education needs for different professions and roles

88. Johnson D, Dissanayake V, Korf BR, Towery M, Haspel RL. An international genomics health workforce education priorities assessment. *Personalized Medicine*. 2022;19(4):299-306.
89. Stark Z, Nisselle A, McClaren B, Lynch F, Best S, Long JC, et al. Attitudes of Australian health professionals towards rapid genomic testing in neonatal and paediatric intensive care. *Eur J Hum Genet*. 2019;27(10):1493-501.
90. Hundertmark ME, Waring SC, Stenehjem DD, Macdonald DA, Sperl DJ, Yapel A, et al. Pharmacist's attitudes and knowledge of pharmacogenomics and the factors that may predict future engagement. *Pharma Prac*. 2020;18(3):2008.
91. Connolly JJ, Berner ES, Smith M, Levy S, Terek S, Harr M, et al. Education and electronic medical records and genomics network, challenges, and lessons learned from a large-scale clinical trial using polygenic risk scores. *Genet Med*. 2023;25(9):100906.
92. Tutika R, Bennett J, Abraham J, Snape K, Tatton-Brown K, Kemp Z, et al. Mainstreaming of genomics in oncology: a nationwide survey of the genomics training needs of UK oncologists. *Clin Med (Lond)*. 2023;23(1):9-15.

Establishing required competency levels across the workforce

93. Rahma AT, Elbarazi I, Ali BR, Patrinos GP, Ahmed LA, Al Maskari F. Genomics and pharmacogenomics knowledge, attitude and practice of pharmacists working in United Arab Emirates: Findings from focus group discussions-A qualitative study. *J Pers Med*. 2020;10(3):134.
94. Korf BR, Berry AB, Limson M, Marian AJ, Murray MF, O'Rourke PP, et al. Framework for development of physician competencies in genomic medicine: Report of the Competencies Working Group of the Inter-Society Coordinating Committee for Physician Education in Genomics. *Genet Med*. 2014;16(11):804-9.

95. Goldgar C, Michaud E, Park N, Jenkins J. Physician assistant genomic competencies. *Journal of Physician Assistant Education*. 2016;27(3):110-6.
96. Calzone KA, Stokes L, Peterson C, Badzek L. Update to the essential genomic nursing competencies and outcome indicators. *J Nurs Schol*. 2024;56(5):729-41.
97. Gammal RS, Lee YM, Petry NJ, Iwuchukwu O, Hoffman JM, Kisor DF, et al. Pharmacists leading the way to precision medicine: Updates to the core pharmacist competencies in genomics. *Am J Pharm Educ*. 2022;86(4):8634.
98. Human Genetics Society of Australasia. Core Capabilities in Genetics & Genomics for Medical Graduates 2022 1 April 2022. Available from: <https://www.hgsa.org.au/documents/item/12553>.
99. NHS Health Education England. Competency frameworks 2022 [Available from: <https://www.genomicseducation.hee.nhs.uk/competency-frameworks/>].
100. Skirton H, Lewis C, Kent A, Coviello DA. Genetic education and the challenge of genomic medicine: development of core competences to support preparation of health professionals in Europe. *Eur J Hum Genet*. 2010;18(9):972-7.
101. Coad B, Joeke K, Rudnicka A, Frost A, Openshaw MR, Tatton-Brown K, et al. Evaluation of two Massive Open Online Courses (MOOCs) in genomic variant interpretation for the NHS workforce. *BMC Med Educ*. 2023;23(1):540.
102. Macfadyen LP, Austin J, Cao P, Cheng J, Cremin C, Duong L, et al. A novel online genomic counseling and variant interpretation certificate: Learning design, learning analytics, and evaluation. *J Genet Couns*. 2023;32(6):1280-7.
103. Dickson A, Cone KR, Fortini BK, Goldstein J, Thompson ML, Wilke MVMB, et al. Variant scientists in genomic medicine: Survey of demographics, training, duties, and professional development. *medRxiv*. 2025.
104. Brooksbank C, Brazas MD, Mulder N, Schwartz R, Ras V, Morgan SL, et al. The ISCB competency framework v. 3: a revised and extended standard for bioinformatics education and training. *Bioinformatics Advances*. 2024;4(1):vbae166.

Evaluation and coordinated efforts can reduce reinvention

105. Maher F, Nisselle A, Lynch E, Martyn M, Tytherleigh R, Charles T, et al. Genomics education for medical specialists: case-based workshops and blended learning. *Journal of Translational Genetics and Genomics*. 2023;7:94-109.
106. Balcom JR, Ellingson MS, Bowler CA, Richardson DM, Kruisselbrink T, Thomas BC. Development of a flipped learning course to deliver and scale molecular variant evaluation education: A quality improvement initiative. *J Genet Couns*. 2024;33(1):168-78.
107. Bishop M, Miller E, McPherson A, Simpson S, Sutherland S, Seller A. Genomic education at scale: the benefits of massive open online courses for the healthcare workforce. *Front Genet*. 2019;10:1094.
108. McCorkell G, Nisselle A, Halton D, Bouffler SE, Patel C, Christodoulou J, et al. A national education program for rapid genomics in pediatric acute care: Building workforce confidence, competence, and capability. *Genet Med*. 2024;26(10):101224.
109. Bennett RL, Waggoner DJ, Blitzer M. Medical genetics and genomics education: how do we define success? Where do we focus our resources? *Genet Med*. 2017;19:751–3.

110. Talwar D, Tseng TS, Foster M, Xu L, Chen LS. Genetics/genomics education for nongenetic health professionals: a systematic literature review. *Genet Med*. 2017;19(7):725-32.
111. McClaren BJ, King EA, Crellin E, Gaff C, Metcalfe SA, Nisselle A. Development of an evidence-based, theory-informed national survey of physician preparedness for genomic medicine and preferences for genomics continuing education. *Front Genet*. 2020;11:59.
112. Nisselle A, Janinski M, Martyn M, McClaren B, Kaunein N, The Reporting Item Standards in Education and Evaluation of Genomics Expert Group, et al. Ensuring best practice in genomics education and evaluation: Reporting Item Standards in Education and Evaluation of Genomics ('RISE2 Genomics'). *Genet Med*. 2021;23(7):1356-65.
113. Nisselle A, Martyn M, Jordan H, Kaunein N, McEwen A, Patel C, et al. Ensuring best practice in genomic education and evaluation: a program logic approach. *Front Genet*. 2019;10:1057.
114. Nisselle A, Macciocca I, McKenzie F, Vuong H, Dunlop K, McClaren B, et al. Readiness of clinical genetic healthcare professionals to provide genomic medicine: An Australian census. *J Genet Couns*. 2019;28(2):367-77.
115. McClaren BJ, King EA, Crellin E, Gaff C, Metcalfe SA, Nisselle A. Development of an Evidence-Based, Theory-Informed National Survey of Physician Preparedness for Genomic Medicine and Preferences for Genomics Continuing Education. *Front Genet*. 2020;11:59.