Ir Med J; October 2023; Vol 116; No. 9; P856
19th October, 2023

Genetic knowledge, experience and educational needs of paediatric trainees in Ireland

S. Quinn, K. Kavanagh, L. Bradley, J. Kenny, S.A. Lynch, A. Green.

Clinical Genetics Department, CHI at Crumlin, Dublin 12, Ireland.

Abstract

Aim

The aim of this study is to assess the current experience and knowledge of genetic testing, consent and interpretation of results amongst paediatric NCHDs working in Children's Health Ireland (CHI).

Methods

A cross-sectional mixed methods survey was distributed to paediatric trainees working within CHI, following the provision of consent from the CHI Ethics Committee. The anonymous survey, designed on Survey Monkey, consisted of 21 questions. It was distributed by email and Whatsapp via lead NCHD and administrative staff.

Results

A total of 32 responses were received, of whom 16 (50%) were senior house officers and 16 (50%) were registrars. 27 (84%) reported they recently consented for genetic testing, with 22 (68%) of those feeling comfortable in doing so. Based on their current knowledge, 13 (41%) felt they had poor understanding of genetic testing technologies and indications, 16 (50%) had a fair understanding of genetic disorders and 17 (53%) had a good or very good knowledge of the consent process. 26 (81%) of NCHDs reported having a fair or good overall understanding of basic genetic science. 26 NCHDs (81%) reported no prior teaching on genetic testing, consent or the implications.

Discussion

The results of this study certainly suggest that paediatricians would benefit from further education on genetic testing and interpretation. The information gathered will assist us in developing effective genetic education strategies to improve the competency of paediatric trainees in this area.

IMJ Official Journal of the Irish Medical Organisation

Ir Med J; October 2023; Vol 116; No. 9; P856
19th October, 2023

Introduction

Genetic testing has become increasingly complex, informative and widely utilised in the world of medicine with human genome sequencing revolutionising our understanding of genetics[1]. In particular, both the consent process and interpretation of genetic testing has become more challenging[2]. Consent has been considered a key element of contemporary medical ethics and clinical practice, and particularly, in relation to genetic testing where the presence of a pathogenic variant may impact not only the patient but additional family members[2]. Genetic testing can also present the risk of incidental findings and variants of uncertain significance which need to be anticipated and included in the consent process[3],[4]. In order for non-genetic healthcare professions to be aware of indications for genetic testing and to effectively consent, they must receive appropriate education on the delivery and interpretation of genetic testing. In Ireland, it is the consultant's responsibility to consent for genetic testing. However, many consultants and non-consultant hospital doctors (NCHDs) do not feel that they receive adequate teaching on available testing or the complex issues and implications surrounding these investigations, as they progress through their careers[5,6]. The predicted, and indeed observable, trend of "mainstreaming of genetic testing" means that non-specialist genetic clinicians are ordering an increasingly potent and complex repertoire of genetic investigations. Safe and effective utilisations of these technologies presuppose a reasonable level of fluency with fundamental genetic concepts, skills and practices. This necessitates the clinician developing this skill set to optimise patient outcomes[7,8].

A study by Burke et al. found that many doctors in specialties where genetic testing is likely to be performed, such as paediatrics, cardiology, dermatology and neurology, are inadequately prepared[9]. Many found it difficult to keep up to date with constantly advancing methods of testing, including microarray, karyotyping and the novel next generation sequencing[10,11]. Microarray (ArrayCGH) is one of the most common requested genetic tests which analyses large genes or proteins to identify abnormal gene expression and copy number variants[12]. Karyotyping is used to identify abnormalities in chromosome size, number or structure in a sample of cells. It is a diagnostic tool performed on different cell types such as blood, skin or buccal cells[13]. Molecular genetic testing utilises DNA sequencing to determine the order of DNA and to identify changes in genes and gene variations[14]. Examples of molecular genetic testing includes whole exome, trio-exome and whole genome sequencing. It was found that primary care practitioners gained much greater confidence in genetic investigations following the Genetics Education Project, where non-genetic professionals provided educational sessions and presentations for their peers and colleagues at an organised workshop[15]. This demonstrates that provision of education sessions will play a key role in the genetic health care nationally. Currently, this has been provided by clinical

geneticists but as healthcare progresses and further discoveries are made, the practice of medicine will likely change opening many opportunities for interprofessional learning. Over time, a common minimum standard of competence in genetics may be required and will help us establish an appropriate framework as the demand for specialist genetic services grow[4,16]. There is certainly a need for focused education and training in genetics to enable paediatric NCHDs to incorporate it into their clinical practices and to remain up to date with this rapidly evolving knowledge base[7,17].

Aim

This study will aim to establish the current experience and knowledge of paediatric NCHDs working in Children's Health Ireland (CHI) services regarding genetic testing, consent and interpretation of results. The purpose of this study will be (1) to determine if paediatric NCHDs have received any education on genetic testing and interpretation to date (2) determine their experience of genomics in the hospital and (3) to explore their comfort level and knowledge of the genetic testing process.

Methods

An online survey and participant information leaflet were distributed to paediatric trainees working in the Children's Health Ireland (CHI) hospitals in November 2022. Consent was obtained from the CHI Ethics Committee. The methodology includes a cross-sectional mixed methods survey using a previously validated questionnaire. The survey incorporates both quantitative and qualitative elements to determine the paediatric NCHDs current experience, education and knowledge of genetic testing processes. A number of the questions aim to establish the frequency of which NCHDs consent for, interpret and communicate genetic results and their comfort levels in doing so. It will aspire to identify the needs and recommendations of paediatric NCHDs regarding genetic testing to improve future educational strategies. The anonymous survey consisted of 21 questions, designed on Survey Monkey in conjunction with clinical genetics specialists to ensure information was appropriate. It was distributed by email and a safe contact platform such as WhatsApp via the lead NCHD and administrative staff.

IMJ Official Journal of the Irish Medical Organisation

Ir Med J; October 2023; Vol 116; No. 9; P856
19th October, 2023

Results

The survey was distributed to 68 paediatric NCHDs working within the tertiary Children's Health Ireland Hospitals (Temple Street Hospital and Our Lady's' Children's Hospital Crumlin). There was a total of 32 responses completed via Survey Monkey. Of those who responded, 16 (50%) were senior house officers and 16 (50%) were registrars working within CHI hospitals. 27 (84%) reported they recently consented for genetic testing, with 22 (68%) of those feeling comfortable in doing so. Based on their current knowledge, 13 (41%) felt they had poor understanding of genetic testing technologies and indications, 16 (50%) had a fair understanding of genetic disorders and 17 (53%) had a good or very good knowledge of the consent process. 26 (81%) of NCHDs reported having a fair or good understanding of basic genetic science and of those 24 (92%), correctly answered the definition questions for microarray, karyotype and molecular genetic testing. An overwhelming 26 NCHDs (81%) have never had teaching on genetic testing, consent or implications. In response to the clinical genetic testing questions, there was an approximate 80% correct response overall. A frequently asked question to the genetics department was regarding the appropriate blood bottles for each test, a simple yet essential step. The survey found that only 14 (43%) were aware of the correct bottle for karyotype, 19 (59%) for ArrayCGH and 18 (56%) for molecular genetic analysis. Of the 19 NCHDs (59%) who felt they were somewhat proficient in genetic testing requesting, 10 (52%) were not aware of the correct bottle required in order for the test to proceed. 23 (71%) reported having fair or good genetic interpretation skills with only 15 (46%) feeling comfortable in communicating these results to patients.

Discussion

The results of this study certainly suggest that paediatricians and paediatric trainees would benefit from further education on genetic testing. The information gathered will aim to assist us in developing effective genetic education strategies and measures to improve paediatric trainees' competencies in this area. In particular, there was a general consensus that enhanced knowledge is required in the areas of genetic testing and technologies, the consent process and interpretation and communication of results. There was adequate expertise on the most frequently utilised genetic tests including microarray, karyotype and molecular genetics. However, it is essential for paediatricians to have a further in-depth awareness of these tests to ensure the most appropriate investigation is requested. Further consideration will be required to determine different strategies for providing effective education and incorporating the opinions and advice of our colleagues will prove invaluable in doing so. As genetic testing becomes increasingly utilised in our daily practice, there will be heightened requirements and expectations for paediatricians to be able to order, interpret and

communicate positive results. We must develop effective methods to tackle this educational gap by working alongside other specialities to devise a pathway and provide appropriate resources in order to guide clinical genetics training for NCHDs and consultants in Ireland.

Declarations of Conflicts of Interest:
None declared.

Corresponding author:
Shauna Quinn,
Clinical Genetics Department,
CHI at Crumlin,
Dublin 12,
Ireland.
E-Mail: squinn8@tcd.ie

References:

1. Genetic education and the challenge of genomic medicine: development of core competences to support preparation of health professionals in Europe
2. Samuel GN, Dheensa S, Farsides B, Fenwick A, Lucassen A. Healthcare professionals' and patients' perspectives on consent to clinical genetic testing: Moving towards a more relational approach [Internet]. BMC medical ethics. BioMed Central; 2017 [cited 2022Nov5].
   Available from: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5549302/
3. Keku TO, Rakhra-Burris T, Millikan R. Gene testing: What the health professional needs to know [Internet]. OUP Academic. Oxford University Press; 2003 [cited 2022Nov5].
   Available from: https://academic.oup.com/jn/article/133/11/3754S/4817983
4. Campion M;Goldgar C;Hopkin RJ;Prows CA;Dasgupta S; Genomic education for the next generation of health-care providers [Internet]. Genetics in medicine : official journal of the American College of Medical Genetics. U.S. National Library of Medicine; [cited 2022Nov5]. Available from: https://pubmed.ncbi.nlm.nih.gov/31110330/
5. Tognetto A, Michelazzo MB, Ricciardi W, Federici A, Boccia S. Core competencies in genetics for healthcare professionals: Results from a literature review and a delphi method [Internet]. BMC medical education. BioMed Central; 2019 [cited 2022Nov5].
   Available from: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6329113/
6. Rosso A;Pitini E;D'Andrea E;Di Marco M;Unim B;Baccolini V;De Vito C;Marzuillo C;Barnhoorn F;Zeegers Paget D;Villari P; Genomics knowledge and attitudes among

European public health professionals: Results of a cross-sectional survey [Internet]. PloS one. U.S. National Library of Medicine; [cited 2022Nov5]. Available from: https://pubmed.ncbi.nlm.nih.gov/32240206/

7. McCarthy J, Patrinos GP. Educating healthcare providers in the delivery of Genomic Medicine. Personalized Medicine. 2019;16(3):187–8.
8. Li J, Xu T, Yashar BM. Genetics educational needs in China: Physicians' experience and knowledge of genetic testing. Genetics in Medicine. 2015;17(9):757–60.
9. Burke S, Stone A, Bedward J, Thomas H, Farndon P. A "neglected part of the curriculum" or "of limited use"? Views on genetics training by nongenetics medical trainees and implications for delivery. Genetics in Medicine. 2006;8(2):109–15.
10. Harding B, Webber C, Rühland L, Dalgarno N, Armour C, Birtwhistle R, et al. Bridging the gap in Genetics: A Progressive model for primary to specialist care. BMC Medical Education. 2019;19(1).
11. Talwar D, Tseng T-S, Foster M, Xu L, Chen L-S. Genetics/Genomics Education for Nongenetic Health Professionals: A systematic literature review. Genetics in Medicine. 2017;19(7):725–32.
12. Kaliyappan K, Govindarajan R, Duraiyan J, Palanisamy M. Microarray and its applications. Journal of Pharmacy And Bioallied Sciences. 2012;4(6):310. doi:10.4103/0975-7406.100283
13. Karyotype [Internet]. [cited 2023 May 15]. Available from: https://www.genome.gov/genetics-glossary/Karyotype
14. [Internet]. [cited 2023 May 15]. Available from: https://www.ncbi.nlm.nih.gov/books/NBK560712/
15. Carroll JC, Rideout AL, Wilson BJ, Allanson JM, Blaine SM, Esplen MJ, Farrell SA, Graham GE, MacKenzie J, Meschino W, Miller F, Prakash P, Shuman C, Summers A, Taylor S. Genetic education for primary care providers: improving attitudes, knowledge, and confidence. Can Fam Physician. 2009 Dec;55(12):e92-9. PMID: 20008584; PMCID: PMC2793208.
16. Calabrò GE, Tognetto A, Mazzaccara A, Barbina D, Carbone P, Guerrera D, et al. Capacity building of health professionals on genetics and Genomics Practice: Evaluation of the effectiveness of a distance learning training course for Italian physicians. Frontiers in Genetics. 2021;12.
17. Rahawi S, Naik H, Blake KV, Owusu Obeng A, Wasserman RM, Seki Y, et al. Knowledge and attitudes on pharmacogenetics among pediatricians. Journal of Human Genetics. 2020;65(5):437–44.