Journal of Neurology & Neurophysiology, 2022, Vol. 13, Issue 4, 001-002

Whole-Exome Sequencing's Cost-Effectiveness in Children with Increasing Neurological Problems

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Received: 3-April -2022, Manuscript No. jnn-22-62786; **Editor assigned:** 16-April -2022, PreQC No. jnn-22-62786 (PQ); **Reviewed**: 24-April -2022, QC No. jnn-22-62786 (Q); **Revised**: 27- April -2022, Manuscript No. jnn-22-62786 (R); **Published**: 29- April -2022, DOI No. 10.35248/2155-9562.2022.13.4.579

Abstract

The purpose of this study was to determine the diagnostic efficacy and cost-effectiveness of whole-exome sequencing (WES) as a standard early-diagnostic tool in children with progressing neurological diseases. During the years 2016–2018, patients with infantile-onset severe neurological illnesses or childhood-onset progressive neurological disorders were prospectively enrolled in the WES study at Helsinki University Hospital's paediatric neurology clinic. A total of 48 individuals underwent a singleton WES. A control group of 49 youngsters had standard diagnostic exams and their results were gathered retrospectively from hospital records. We acquired information about their usage of healthcare services relevant to the diagnosis procedure. From the standpoint of the healthcare provider, the Incremental Cost-Effectiveness Ratio (ICER) per extra diagnostic was determined. The uncertainty of cost-effectiveness results was estimated using bootstrapping methods. WES had a 38% higher diagnostic yield than a diagnostic route that did not emphasize WES in early diagnosis. WES outperformed other diagnostic approaches, particularly when conducted early, within a year of admission. Our cost-effectiveness estimates are cautious, as they are influenced by WES expenses from 2016 to 2018.

Keywords: Diagnosis • Whole-exome sequencing

Introduction

Over 4000 clinical synopses with neurological involvement are recognized in the Online Mendelian Inheritance of Man database, with over 3000 having a verified molecular foundation. The genetic complexity is significant: a single variant can cause variable symptoms in different patients, as in X-linked adrenoleukodystrophy. For example, Leigh's disease can be caused by variants in more than 75 different genes, and even a single variant can cause variable symptoms in different patients. The development of next-generation sequencing (NGS) tools for the human genome has greatly enhanced diagnostic procedures and in certain cases, has provided focused therapeutic options. Furthermore, early genetic diagnosis gives counselling and reproductive planning information. Whole-exome sequencing (WES) extracts information from the genome's protein-coding genes. When all genes are sequenced instead of just a few candidate genes, the time it takes to find gene abnormalities is cut in half, and new disease-causing genes can be discovered. The cost of analysis has also decreased as a result of method improvement, making regular diagnostics possible. However, health care providers may still perceive NGS technologies to be too costly for clinical use, necessitating cost-effectiveness studies to aid decision-making [1]. Better health outcomes or more effective utilization of health care services may arise from the enhanced and efficient diagnostic output as a result of NGS analysis.

According to a recent meta-analysis, WES has a pooled diagnostic value of 36% in children with suspected genetic disorders, which means the rate of conclusive diagnoses reached. However, there is just a little research on the cost-effectiveness and economic effects of WES currently available. The expenses of genetic tests and WES are shown to be the main cost drivers in a diagnostic work-up to achieve a diagnosis, but if WES is employed as a near-first-line test in a selected cohort of patients, and overall budget increase may not be necessary[2].

The diagnostic value and cost-effectiveness of WES as a routine diagnostic tool in paediatric patients with progressing neurological diseases are assessed in this study. Our findings suggest that WES has a higher diagnosis yield (37.5 vs. 24.5%) than a traditional diagnostic strategy using clinical diagnostic methods and gene panel testing. The "early-WES" programme in the first year was undoubtedly the most successful. Our WES group's diagnostic yield matches a newly published meta-analysis of children with suspected genetic disorders. WES led to previously unattainable diagnoses for four out of fifteen patients who were recruited to the trial even after three years of earlier investigations (31%), who had been evaluated using a comprehensive variety of traditional diagnostic techniques [3].

The study's strength is its prospective cohort study design, which allowed researchers to investigate WES as a regular diagnostic tool. A retrospectively collected control group of patients who underwent standard diagnostic testing is also included in the research. However, there are several limitations to this research. The sample size is rather small, and the dwelling district may provide a selection bias. Diagnostic yield varies depending on how effectively the initial patient sample was preselected in various research, and this has a direct impact on cost-effectiveness. If trio-analysis had been used, the diagnostic yield would have been higher. However, in WES studies, yields of 30 to 40% are frequently attained, demonstrating the diagnostic tool's utility. Diagnostic yield for singleton-WES was found to be 26.5% across studies in a benchmark meta-study of children with varied suspected genetic disorders, indicating that this range of yield is typical for child presentations. The sample size of our investigation, which was limited due to a lack of financial resources to conduct WES, may have extended the bootstrapped results' confidence ranges. Third, because the study's goal was to determine the costs of early WES analysis, this sample was chosen at random. Finally, infantile encephalopathies and progressive neurological illnesses of infancy are clinically diverse groups of patients, and to obtain a precise diagnosis, a marathon of investigations is frequently undertaken, raising non-WES diagnostic expenditures. Similar cost-effectiveness analyses for various patient categories might be beneficial [4].

The findings are particularly intriguing because our research group was clinically and genetically varied and had a wide clinical definition - a progressive neurological disease of infancy. We recommend that WES be employed in the first-line diagnosis of undefined progressive neurological illnesses in children because a third of these patients would receive a diagnosis immediately, and therapy could be tailored to the disease. The advancement of NGS technologies and analysis, as well as the steadily dropping cost of WES, make the procedure extremely helpful in the diagnosis of children. Future research should include societal economic evaluations, including expenditures after WES; a recent article found that diagnosis-related physician consultations do not decrease after a bad WES. Furthermore, the cost-effectiveness should be evaluated using more generic efficacy indicators, such as Quality Adjusted Life Years (QALYs) [5].

Conclusion

WES is a cost-effective and efficient diagnostic technique that should be used in the early stages of the diagnosis of children with progressing neurological diseases. The test's cost-effectiveness is further enhanced by its steadily reducing price.

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Cite This Article: Williams C. Whole-Exome Sequencing's Cost-Effectiveness in Children with Increasing Neurological problems. Neurol Neurophysiol. 2022,13(4), 001-002.