**Perspective**
Volume 8:2, 2023

Journal of Pediatric Neurology and Medicine

ISSN: 2472-100X

Open Access

# Whole-exome Sequencing's Value in Children with Increasing Neurological Problems

**Lieke Ghanim***

*Department of Paediatrics, University of Medical Center Groningen, Groningen, The Netherlands*

## Introduction

Over 4000 clinical synopses with neurological involvement are identified by the Online Mendelian Inheritance of Man database, of which over 3000 has a demonstrated molecular basis. The significance of genetic complexity lies in: The same disease can be caused by mutations in multiple genes; for instance, Leigh disease can be caused by mutations in more than 75 genes. Even a single mutation can cause X-linked adrenoleukodystrophy to have distinct symptoms in different patients. The advancement of cutting edge sequencing (NGS) strategies for the human genome emphatically further developed the indicative methodologies in some cases giving designated ways to deal with treatment. Additionally, early genetic diagnosis provides counseling and reproductive planning advice and tools [1].

## Description

The genome's protein-coding genes are the source of the data provided by whole-exome sequencing (WES). When compared to single candidate genes, sequence analysis of all genes speeds up the process of identifying gene defects and identifies novel disease-causing genes. Additionally, the development of methods has gradually reduced the cost of the analysis, making routine diagnostics possible. However, healthcare providers may still consider NGS methods to be too costly for clinical practice, necessitating cost-effectiveness studies to aid in decision-making. Better health outcomes or a more effective use of health care services might result from the increased diagnostic yield that NGS-analysis produces. A recent meta-analysis revealed that in children with suspected genetic diseases, WES had a pooled diagnostic utility of 36%, or the rate at which definitive diagnoses were made. However, there are only a few studies currently available on WES's cost-effectiveness and economic outcomes [2].

The costs of genetic tests and WES have been identified as the primary cost drivers in diagnostic workup, but if WES is used as a near-first-line test in a select group of patients, an overall budget increase may not be necessary. Patients with childish beginning extreme neurological illness or youth beginning moderate neurological issue were tentatively selected to the WES learn at Kids' Emergency clinic at Helsinki College Medical clinic, a tertiary consideration medical clinic, during the years 2016-2018. Non-progressive intellectual disability or autism spectrum disorder, a family history of a known genetic disorder, or any other genetic disorder that could be clinically identified were the exclusion criteria. As a routine diagnostic procedure, the singleton WES was administered to 48 non-consanguineous pediatric patients (the "WES group").

The study participants' utilization of health care services related to their diagnostic path was gathered retrospectively from patient records. All diagnostic health care visits and investigations—hospitalizations, clinical visits, laboratory tests, imaging, and genetic testing—were included in the data. The events were individually reviewed by the study physicians, and only those that were deemed pertinent to the diagnostic process were included. What's more, orientation and age at the primary visit in the medical clinic, the date of analysis and timing of WES along the demonstrative way were recorded [3].

The hospital as a health care provider was used in the economic analysis. The hospital (Hospital District of Helsinki and Uusimaa, HUS) and diagnostic laboratory documentation were used to obtain costs for genetic, imaging, and laboratory tests. The hospital district's outpatient product costs for specialized somatic health care visits served as the basis for defining clinical visit costs. The expenses for hospitalization not entirely settled from the assessments by Finnish Public Foundation for Wellbeing and Government assistance for the unit expenses of social and medical services in 2011. The expenses of non-WES symptomatic tests in 2019 were changed over completely to 2018 costs in euros utilizing the public wellbeing and social consideration cost record by the Relationship of Finnish Nearby and Provincial Specialists and money converter or the ongoing cost was utilized, for example for demonstrative tests performed external the emergency clinic. According to the commercial price used in Helsinki hospital district's laboratory (HUSLAB) in November 2019, the WES price, which includes all technical and analytical costs as well as staff salaries, was estimated to be 1375€ per singleton WES [4].

Cross-tabulation, chi-square, and Fisher's exact tests were used to compare the baseline characteristics of the children in the WES group and the control group. The Wilcoxon rank-sum test was used to analyze continuous variables. Standard deviations, medians, and 95 percent confidence intervals (CI) were used to calculate the mean diagnostic costs for each patient. Additionally, total costs were divided by the total number of diagnoses in the groups to determine mean costs per diagnosis.

In pediatric patients with progressive neurological disorders, the diagnostic utility and cost-effectiveness of WES as a routine diagnostic tool are the focus of this investigation. According to our findings, compared to the conventional diagnostic approach that employs clinical diagnostic methods supplemented with gene panel testing, WES yields a higher diagnostic yield (37.5% versus 24.5%). It was evident that first-year "early-WES" students performed the best. A meta-analysis of children with suspected genetic diseases that was recently published is consistent with our diagnostic yield in the WES group. WES resulted in previously unattainable diagnoses for four out of fifteen patients, taking into account patients who were recruited to the study even after three years of prior investigations (31%) and had been examined with a large set of standard diagnostic tools.

To clarify the total costs of the examinations, we decided to collect the WES and conventional diagnostic path costs in full. Patients from a similar control group have not been used in previous studies. Diagnostic scenarios in the same study cohort or a hypothetical WES trajectory were used to model a lot of previous studies. In addition, Europe was the location of only a few studies. Additionally, prior research focused primarily on the cost-effectiveness of WES in pediatric patients with any suspected monogenic disorders or specific disorders, such as epilepsy or muscle disorders. In accordance

***Address for Correspondence:** Lieke Ghanim, Department of Paediatrics, University of Medical Center Groningen, Groningen, The Netherlands, E-mail: liekeghanim@gmail.com*



**Received:** 01 March 2023, Manuscript No. JPNM-23-95335; **Editor assigned:** 03 March 2023, Pre QC No. P-95335; **Reviewed:** 15 March 2023, QC No. Q-95335; **Revised:** 20 March 2023, Manuscript No. R-95335; **Published:** 27 March 2023, DOI: 10.37421/2472-100X.2023.8.220

with previous research conducted on pediatric cohorts and mixed cohorts of children and adults with complex neurological problems, the finding that clinical visits and genetic tests were the primary drivers of costs in both study groups is consistent [5].

## Conclusion

It was mentioned that health status or quality of life might not be the only outcome measure in health economic evaluations of genetic testing. This is because genetic information is valuable and can affect one's ability to make informed decisions. The payer's willingness to pay for one additional diagnosis determines cost-effectiveness, as there is no single threshold for interpreting our study's ICER result. Estimating such willingness to pay and determining whether payers are eager to reimburse on such outcome measures require additional research. The significance of genetic testing cannot be overstated because it eliminates the need for diagnostic tests, provides precise genetic counseling and diagnosis, provides prognosis, and occasionally directs treatment decisions.

## Acknowledgement

None.

## Conflict of Interest

None.

## References

1. Schwarze, Katharina, James Buchanan, Jenny C. Taylor and Sarah Wordsworth. "Are whole-exome and whole-genome sequencing approaches cost-effective? A systematic review of the literature." *Genet Med* 20 (2018): 1122-1130.
2. Grosse, Scott D, Sarah Wordsworth and Katherine Payne. "Economic methods for valuing the outcomes of genetic testing: Beyond cost-effectiveness analysis." *Genet Med* 10 (2008): 648-654.
3. Stark, Zornitza, Deborah Schofield, Melissa Martyn and Luke Rynehart, et al. "Does genomic sequencing early in the diagnostic trajectory make a difference? A follow-up study of clinical outcomes and cost-effectiveness." *Genet Med* 21 (2019): 173-180.
4. Van Nimwegen, Kirsten JM, Ronald A. van Soest and Joris A. Veltman, et al. "Is the $1000 genome as near as we think? A cost analysis of next-generation sequencing." *Clin Chem* 62 (2016): 1458-1464.
5. Sainio, Markus T, Salla Välipakka, Bruno Rinaldi and Helena Lapatto, et al. "Recessive pyroxd1 mutations cause adult-onset limb-girdle-type muscular dystrophy." *J Neurol* 266 (2019): 353-360.

**How to cite this article:** Ghanim, Lieke. "Whole-exome Sequencing's Value in Children With Increasing Neurological Problems." *J Pediatr Neurol Med* 8 (2023): 220.