ARE WHOLE EXOME AND WHOLE GENOME SEQUENCING APPROACHES COST-EFFECTIVE? A SYSTEMATIC REVIEW OF THE LITERATURE

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OBJECTIVES: Evidence suggests that next generation sequencing technologies could improve the diagnosis and treatment of genetic diseases. However, demand is increasing for evidence on the costs and health outcomes associated with these technologies. Our objective was to conduct a systematic literature review to summarise the current health economic evidence for the use of whole exome sequencing (WES) and whole genome sequencing (WGS) in clinical settings. **METHODS:** Relevant studies were identified in the EMBASE, MEDLINE, Cochrane Library, EconLit and University of York Centre for Reviews and Dissemination databases from January 2005 to July 2016. Publications were included in the review if they were economic evaluations, cost studies or outcome studies. Data were extracted from each publication on sample size, sequencing methods, study methods, outcomes, costs and main results. **RESULTS:** Thirty-six studies met our inclusion criteria (21 economic evaluations, 7 cost studies, 8 outcome studies). These publications investigated the use of WES and WGS in a variety of genetic conditions, the most common being neurological or neurodevelopmental disorders. Study sample size varied from a single child to 2,000 patients. Cost estimates for a single test ranged from $555-$5,169 for WES and from $1,906-$24,810 for WGS. There was no evidence that the cost of WES was falling over time, and only limited evidence that the cost of WGS was reducing. Few cost analyses presented data transparently or stated which components were included in cost estimates. In addition, few studies used outcome measures recommended for use in economic evaluations, such as survival or quality of life. **CONCLUSIONS:** The current health economic evidence base to support the more widespread use of WES and WGS in clinical practice is very limited. Studies that carefully evaluate the costs, health outcomes and cost-effectiveness of these tests are urgently needed to support their translation into clinical practice.

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