

References

L-206

1. Shashi V, McConkie-Rosell A, Rosell B, et al. The utility of the traditional medical genetics diagnostic evaluation in the context of next-generation sequencing for undiagnosed genetic disorders. *Genet Med*. 2014 Feb; 16: 176–182. PMID: 23928913.
2. Sawyer SL, Hartley T, Dymont DA et al. Utility of whole-exome sequencing for those near the end of the diagnostic odyssey: time to address gaps in care. *Clin Genet*. 2016 Mar; 89: 275–284. PMID:26283276.
3. Dixon-Salazar TJ, Silhavy JL, Udpa N, et al. Exome sequencing can improve diagnosis and alter patient management. *Sci Transl Med*. Jun 13 2012;4(138):138ra178. PMID 22700954.
4. Halverson CM, Clift KE, & JB McCormick. Was it worth it? Patients' perspectives on the perceived value of genomic-based individualized medicine. *J Community Genet*. Apr 2016; 7(2):145–152. PMID 26860291.
5. Iglesias A, Anyane-Yeboah K, Wynn J, et al. The usefulness of whole-exome sequencing in routine clinical practice. *Genet Med*. Dec 2014;16(12):922-931. PMID 24901346.
6. Nolan D, Carlson M. Whole exome sequencing in pediatric neurology patients: clinical implications and estimated cost analysis. *J Child Neurol*. Jun 2016;31(7):887-894. PMID 26863999.
7. Stark Z, Tan TY, Chong B, et al. A prospective evaluation of whole-exome sequencing as a first-tier molecular test in infants with suspected monogenic disorders. *Genet Med*. Nov 2016;18(11):1090-1096. PMID 26938784.
8. Thevenon J, Duffourd Y, Masurel-Paulet A, et al. Diagnostic odyssey in severe neurodevelopmental disorders: toward clinical whole-exome sequencing as a first-line diagnostic test. *Clinical Genetics*. 2016 Jun; 89: 700–707. PMID: 26757139.
9. Valencia CA, Husami A, Holle J, et al. Clinical impact and cost-effectiveness of whole exome sequencing as a diagnostic tool: a pediatric center's experience. *Front Pediatr*. 2015;3:67 PMID 26284228.
10. Rehm HL, Bale SJ, Bayrak-Toydemir P, et al. ACMG clinical laboratory standards for nextgeneration sequencing. *Genet Med*. Sep 2013;15(9):733-747. PMID 23887774.
11. ACMG Board of Directors. Points to consider in the clinical application of whole-genome sequencing. *Genet Med*. 2012; 14:759–761. PMID 22863877.
12. ACMG Board of Directors. Points to consider for informed consent for genome/exome sequencing. *Genet Med*. Sept 2013;15(9):748–749. PMID 23970068.
13. Green RC, Berg JS, Grody WW, et al. ACMG recommendations for reporting of incidental findings in clinical exome and genome sequencing. *Genet Med*. Jul 2013;15(7):565-574. PMID 23788249.
14. Kalia SS, Adelman K, Bale SJ, et al. Recommendations for reporting of secondary findings in clinical exome and genome sequencing, 2016 update (ACMG SF v2.0): a policy statement of the American College of Medical Genetics and Genomics. *Genet Med*. 2016 Nov 17. doi: 10.1038/gim.2016.190. [Epub ahead of print] PMID: 27854360.

15. Farwell KD, Shahmirzadi L, El-Khechen D, et al. Enhanced utility of family-centered diagnostic exome sequencing with inheritance model-based analysis: results from 500 unselected families with undiagnosed genetic conditions. *Genet Med*. Jul 2015;17(7):578- 586. PMID 25356970.
16. Yang Y, Muzny DM, Xia F, et al. Molecular Findings Among Patients Referred for Clinical Whole-Exome Sequencing. *JAMA*. 2014;312(18):1870-1879. PMID 25326635.
17. Trujillano D, Bertoli-Avella AM, Kumar Kandaswamy K, et al. Clinical exome sequencing: results from 2819 samples reflecting 1000 families. *Eur J Hum Genet*. 2017 Feb;25(2):176- 182. PMID 27848944.
18. Lee H, Deignan JL, Dorrani N, et al. Clinical exome sequencing for genetic identification of rare Mendelian disorders. *JAMA*. Nov 12 2014;312(18):1880-1887. PMID 25326637.
19. Retterer K, Juusola J, Cho MT, et al. Clinical application of whole-exome sequencing across clinical indications. *Genet Med*. Dec 2016; 18: 696–704. PMID 26633542.
20. Wenger AM, Guturu H, Bernstein JA, et al. Systematic reanalysis of clinical exome data yields additional diagnoses: implications for providers. *Genet Med*. Jul 2016. Epub, ahead of print. PMID 27441994.