

## ***Ethical Dilemmas of Genomic Sequencing***

### References

- Allanson JE, Roberts AE. Noonan Syndrome. 2001 Nov 15 [updated 2019 Aug 8]. In: Adam MP, Ardinger HH, Pagon RA, Wallace SE, Bean LJH, Stephens K, Amemiya A, editors. GeneReviews<sup>®</sup> [Internet]. Seattle (WA): University of Washington, Seattle; 1993–2020. PMID: 20301303.
- Berg JS, Agrawal PB, Bailey DB Jr, Beggs AH, Brenner SE, et al. Newborn Sequencing in Genomic Medicine and Public Health. *Pediatrics*. 2017 Feb;139(2):e20162252. doi: 10.1542/peds.2016-2252. Epub 2017 Jan 17. PMID: 28096516; PMCID: PMC5260149.
- Berg JS. Genome-scale sequencing in clinical care: establishing molecular diagnoses and measuring value. *JAMA*. 2014 Nov 12;312(18):1865-7. doi: 10.1001/jama.2014.14665. PMID: 25326641.
- Biesecker LG. Opportunities and challenges for the integration of massively parallel genomic sequencing into clinical practice: lessons from the ClinSeq project. *Genet Med*. 2012 Apr;14(4):393-8. doi: 10.1038/gim.2011.78. Epub 2012 Feb 16. PMID: 22344227; PMCID: PMC3790899.
- Daoud H, Luco SM, Li R, Bareke E, Beaulieu C, et al. Next-generation sequencing for diagnosis of rare diseases in the neonatal intensive care unit. *CMAJ*. 2016 Aug 9;188(11):E254-E260. doi: 10.1503/cmaj.150823. Epub 2016 May 30. PMID: 27241786; PMCID: PMC4978597.
- Iglesias A, Anyane-Yeboah K, Wynn J, Wilson A, Truitt Cho M, et al. The usefulness of whole-exome sequencing in routine clinical practice. *Genet Med*. 2014 Dec;16(12):922-31. doi: 10.1038/gim.2014.58. Epub 2014 Jun 5. PMID: 24901346.
- Johnston J, Lantos JD, Goldenberg A, Chen F, Parens E, Koenig BA; members of the NSIGHT Ethics and Policy Advisory Board. Sequencing Newborns: A Call for Nuanced Use of Genomic Technologies. *Hastings Cent Rep*. 2018 Jul;48 Suppl 2(Suppl 2):S2-S6. doi: 10.1002/hast.874. PMID: 30133723; PMCID: PMC6901349.
- Kwon JM, Matern D, Kurtzberg J, Wrabetz L, Gelb MH, et al. Consensus guidelines for newborn screening, diagnosis and treatment of infantile Krabbe disease. *Orphanet J Rare Dis*. 2018 Feb 1;13(1):30. doi: 10.1186/s13023-018-0766-x. PMID: 29391017; PMCID: PMC5796396.
- Langan TJ, Orsini JJ, Jalal K, Barczykowski AL, Escolar ML, et al. Development of a newborn screening tool based on bivariate normal limits: using psychosine and galactocerebrosidase determination on dried blood spots to predict Krabbe disease. *Genet Med*. 2019 Jul;21(7):1644-1651. doi: 10.1038/s41436-018-0371-3. Epub 2018 Dec 14. PMID: 30546085.
- Lantos JD. The False-negative Phenotype. *Pediatrics*. 2019 Jan;143(Suppl 1):S33-S36. doi: 10.1542/peds.2018-1099G. PMID: 30600269.
- Lindor NM, Thibodeau SN, Burke W. Whole-Genome Sequencing in Healthy People. *Mayo Clin Proc*. 2017 Jan;92(1):159-172. doi: 10.1016/j.mayocp.2016.10.019. PMID: 28062062.
- Park A. Genetic screening saved this baby's life. *Time Magazine*. Dec 8, 2014. <http://time.com/3616907/genetic-screening-saved-this-babys-life/>

Petrikin JE, Cakici JA, Clark MM, Willig LK, Sweeney NM, et al. The NSIGHT1-randomized controlled trial: rapid whole-genome sequencing for accelerated etiologic diagnosis in critically ill infants. *NPJ Genom Med*. 2018 Feb 9;3:6. doi: 10.1038/s41525-018-0045-8. PMID: 29449963; PMCID: PMC5807510.

Stark Z, Tan TY, Chong B, Brett GR, Yap P, et al. A prospective evaluation of whole-exome sequencing as a first-tier molecular test in infants with suspected monogenic disorders. *Genet Med*. 2016 Nov;18(11):1090-1096. doi: 10.1038/gim.2016.1. Epub 2016 Mar 3. PMID: 26938784.

Surtees R, Wolf N. Treatable neonatal epilepsy. *Arch Dis Child*. 2007 Aug;92(8):659-61. doi: 10.1136/adc.2007.116913. PMID: 17642476; PMCID: PMC2083881.

Thiffault I, Lantos J. The Challenge of Analyzing the Results of Next-Generation Sequencing in Children. *Pediatrics*. 2016 Jan;137 Suppl 1:S3-7. doi: 10.1542/peds.2015-3731C. PMID: 26729700.

Wasserstein MP, Andriola M, Arnold G, Aron A, Duffner P, et al. Clinical outcomes of children with abnormal newborn screening results for Krabbe disease in New York State. *Genet Med*. 2016 Dec;18(12):1235-1243. doi: 10.1038/gim.2016.35. Epub 2016 May 12. PMID: 27171547.

Worthey EA, Mayer AN, Syverson GD, Helbling D, Bonacci BB, et al. Making a definitive diagnosis: successful clinical application of whole exome sequencing in a child with intractable inflammatory bowel disease. *Genet Med*. 2011 Mar;13(3):255-62. doi: 10.1097/GIM.0b013e3182088158. PMID: 21173700.

Willig LK, Petrikin JE, Smith LD, Saunders CJ, Thiffault I, et al. Whole-genome sequencing for identification of Mendelian disorders in critically ill infants: a retrospective analysis of diagnostic and clinical findings. *Lancet Respir Med*. 2015 May;3(5):377-87. doi: 10.1016/S2213-2600(15)00139-3. Epub 2015 Apr 27. PMID: 25937001; PMCID: PMC4479194.

<http://www.latimes.com/local/lanow/la-me-ln-newborn-sequencing-20180422-htlstory.html>