

# Should all babies have their genomes sequenced?

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As the cost of genome sequencing decreases, researchers and clinicians are debating whether all newborns should be sequenced at birth, facilitating a lifetime of personalized medical care. But while sequencing

the genomes of some infants may be appropriate in specific contexts, genome-wide sequencing of all newborns should not be pursued at this time, and health professionals should recommend against parents using direct-to-consumer genetic sequencing to diagnose or screen their newborns, states the lead article in [The Ethics of Sequencing Newborns: Recommendations and Reflections](#), a new special report of the Hastings Center Report.

Josephine Johnston, director of research at The Hastings Center; Erik Parens, senior research scholar at The Hastings Center; and Barbara Koenig, a professor at the University of California, San Francisco, and director of the UCSF Program in Bioethics who is a Hastings Center Fellow, are co-editors of the special report.

The lead article was written by members of the University of California, San Francisco, Newborn Sequencing in Genomic Medicine and Public Health (NSIGHT) Ethics and Policy Advisory Board, composed of researchers and scholars from genomics, clinical medicine, bioethics, and other fields. Their recommendations grew out of a four-year interdisciplinary investigation funded by the Eunice Kennedy Shriver National Institute of Child Health and Human Development and the National Human Genome Research Institute, two components of the National Institutes of Health, to examine the ethical and policy issues posed by conducting genome sequencing on newborns.

"Genomics is a powerful tool, but the results it returns are still not fully understood and have not been proven to advance health outside of very specific clinical situations," says Johnston. "The recommendations embrace the use of genomics to aid in the diagnosis of sick newborns, but they draw a sharp distinction between that kind of focused clinical use and population screening."

The recommendations appear in "[Sequencing Newborns: A Call for](#)

[Nuanced Use of Genomic Technologies.](#)" They include:

- Targeted or [genomic sequencing](#) can be used by clinicians to assist in the diagnosis of a symptomatic newborn. Sequencing these newborns may end the search for a diagnosis, informing medical management.
- Genome-wide sequencing should not be implemented as a universal, public health screening tool in newborns. Sequencing the entire genome may result in the return of genetic data of unknown or uncertain significance and may not yield actionable results. Results can generate unnecessary distress and require health resources for unneeded monitoring. And the cost of universal genome-wide sequencing would stretch the operating expenses of state-funded newborn screening programs, undermining the effectiveness of their operations.
- Integrating targeted genome sequencing into newborn screening programs may be appropriate when it is the best way to identify a condition that meets existing screening criteria—it affects a newborn's health, programs are able to fund screening and follow-up care, and effective treatments are available. Targeted genome sequencing may also be appropriate to confirm a diagnosis and provide additional prognostic information after initial screening results.
- Whole-genome or targeted sequencing should not be integrated into routine infant primary care. In healthy babies, genome sequencing would likely generate undue anxiety and require significant health resources for interpretation and follow-up.
- Health professionals should recommend against parents seeking direct-to-consumer [genome sequencing](#) for either diagnosis or screening of their newborn. The use of DTC genomic testing in children conflicts with clinical and professional guidelines, which limit testing to clinical contexts and for conditions that manifest during childhood. Most testing services also lack sufficient

consultation and follow-up to assure accurate interpretation of results.

"Sequencing the genome of every newborn could cause parents to worry needlessly about their healthy baby," says Koenig.

Twelve essays expand upon the recommendations in the lead article, exploring a range of issues. Among the essays:

["Families' Experiences with Newborn Screening: A Critical Source of Evidence"](#) calls for more research on the impact of expanded newborn screening on the lived experience of parents and children. Such studies "will be essential for guiding decisions about the future," write Rachel Grob, clinical professor in the Department of Family Medicine and Community Health at the University of Wisconsin-Madison; Scott Roberts, associate professor of health behavior and health education at the University of Michigan School of Public Health; and Stefan Timmermans, professor of sociology at the University of California, Los Angeles.

["Commercial Interests, the Technological Imperative, and Advocates: Three Forces Driving Genomic Sequencing in Newborns"](#) raises concern about forces, "beyond the desire to implement tests with proven clinical utility, that are fueling interest in genomic sequencing in the newborn period. These three forces have the potential to be problematic for policy and practice." The authors are Stacey Pereira, an assistant professor at the Center for Medical Ethics and Health Policy at Baylor College of Medicine, and Ellen Wright Clayton, a professor of pediatrics, law and [health](#) policy at Vanderbilt University.

["Using Newborn Sequencing to Advance Understanding of the Natural History of Disease"](#) argues that genomic sequencing of sick newborns has the potential to bypass the prolonged journey to a diagnosis,

improving the medical care of individual infants. "But sequencing also has the potential to benefit others beyond the child whose [genome](#) is sequenced and his or her immediate family. Sequence data from sick newborns will expand medicine's understanding of genetic diseases," writes Ingrid A. Holm, an associate professor of pediatrics at Harvard Medical School.

The entire report is available for free [here](#).

Provided by The Hastings Center

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