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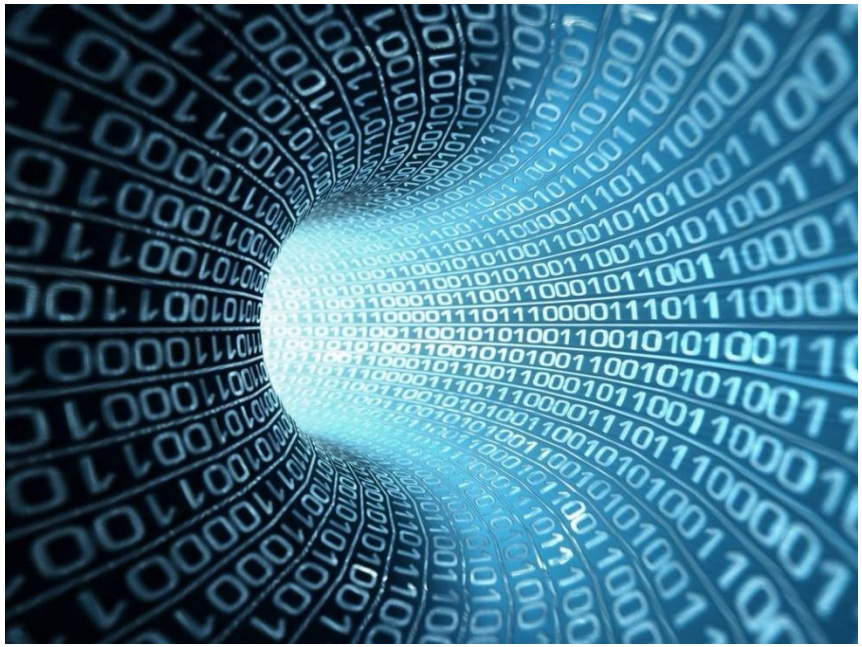
BIG Data



- Journals are traditionally one of the main repositories of data.
- Datasets are increasing in size and complexity.
- Primary concern is access.
  - Print is not sufficient.
  - Supplemental materials
  - Deposit in accessible database.

Approved Databases

Molecular structure data	Worldwide Protein Data Bank; BioMag Res Bank; Electron Microscopy Data Bank; Cambridge Crystallographic Data Centre
DNA and protein sequences	GeneBank; EMBL; DDBJ; SWISS-PROT
Microarray data	Gene Expression Omnibus; ArrayExpress

GENOMICS

Noninvasive Whole-Genome Sequencing of a Human Fetus

Jacob O. Kitzman,[1]* Matthew W. Snyder,[1] Mario Ventura,[1,2] Alexandra P. Lewis,[1] Ruolan Qiu,[1] LaVone E. Simmons,[3] Hilary S. Gammill,[3,4] Craig E. Rubens,[5,6] Donna A. Santillan,[7] Jeffrey C. Murray,[8] Holly K. Tabor,[5,9] Michael J. Bamshad,[1,5] Evan E. Eichler,[1,10] Jay Shendure[1]*

Analysis of cell-free fetal DNA in maternal plasma holds promise for the development of noninvasive prenatal genetic diagnostics. Previous studies have been restricted to detection of fetal trisomies, to specific paternally inherited mutations, or to genotyping common polymorphisms using material obtained invasively, for example, through chorionic villus sampling. Here, we combine genome sequencing of two parents, genome-wide maternal haplotyping, and deep sequencing of maternal plasma DNA to noninvasively determine the genome sequence of a human fetus at 18.5 weeks of gestation. Inheritance was predicted at 2.8×10^6 parental heterozygous sites with 98.1% accuracy. Furthermore, 39 of 44 de novo point mutations in the fetal genome were detected, albeit with limited specificity. Subsampling these data and analyzing a second family trio by the same approach indicate that parental haplotype blocks of ~300 kilo–base pairs combined with shallow sequencing of maternal plasma DNA is sufficient to substantially determine the inherited complement of a fetal genome. However, ultradeep sequencing of maternal plasma DNA is necessary for the practical detection of fetal de novo mutations genome-wide. Although technical and analytical challenges remain, we anticipate that noninvasive analysis of inherited variation and de novo mutations in fetal genomes will facilitate prenatal diagnosis of both recessive and dominant Mendelian disorders.

61/651,356). **Data and materials availability:** The data for this study have been deposited in the database dbGaP under accession "phs000500.v1.p1."

dbGaP phs000500.v1.p1

Search results: 19 Variables, 0 Analyses, 0 Documents, and 4 Datasets in 1 Studies

Studies (1) | **Variables (19)** | **Study Documents (0)** | **Analyses (0)** | **Datasets (4)**

Study	Embargo Release	Details	Participants	Type Of Study	Links	Platform
phs000500.v1.p1 Non-Invasive Whole Genome Sequencing of a Human Fetus	Version 1: passed embargo	V D A S	6	Parent-Offspring Trios	Links	

Data Storage

- Searchable
- Centralized
- Well-maintained
- Challenges –
  - Cross-referencing different types of data from different sites.
  - Quality control

Enforcement

- Site maintenance
- Ensuring public access
- Individual privacy
- Errors
- Data sharing