

Exploring the Current Landscape of Consumer Genomics

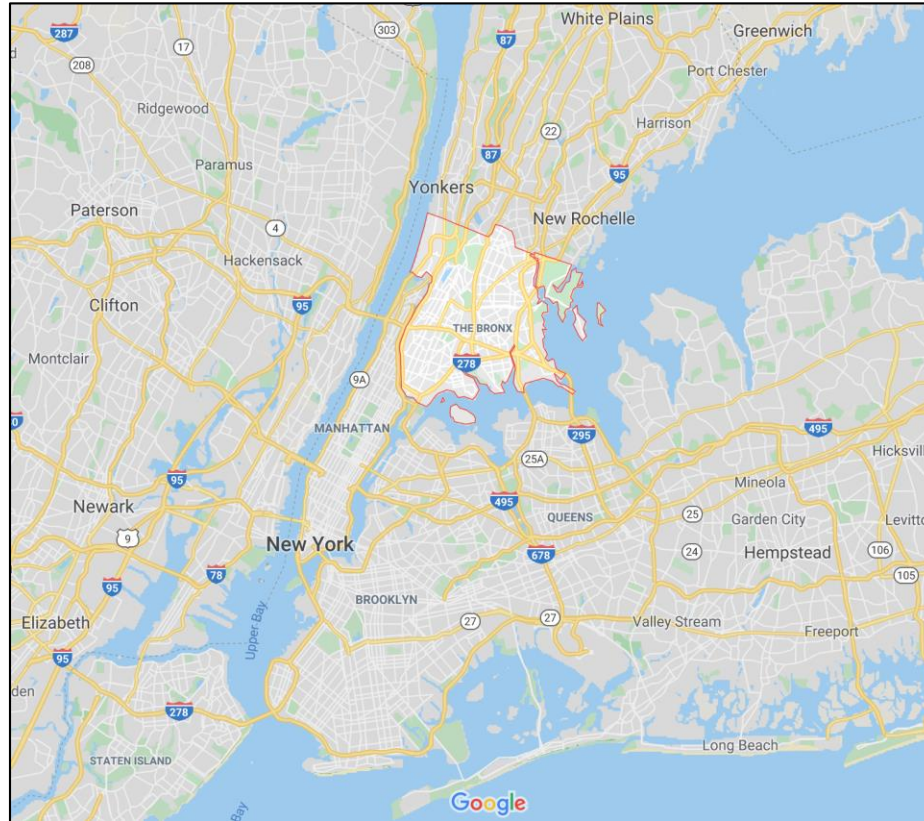
Siobhan Dolan, MD, MPH
Professor and Vice Chair, Research

Department of Obstetrics & Gynecology and Women's Health
Division of Reproductive and Medical Genetics
Albert Einstein College of Medicine
Montefiore Health System
Bronx, NY

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The Bronx, NYC



The Bronx, NYC

Population estimates, July 1, 2018, (V2018)

1,432,132

Race and Hispanic Origin	
i White alone, percent	⚠ 44.9%
i Black or African American alone, percent (a)	⚠ 43.6%
i American Indian and Alaska Native alone, percent (a)	⚠ 2.9%
i Asian alone, percent (a)	⚠ 4.5%
i Native Hawaiian and Other Pacific Islander alone, percent (a)	⚠ 0.4%
i Two or More Races, percent	⚠ 3.7%
i Hispanic or Latino, percent (b)	⚠ 56.4%
i White alone, not Hispanic or Latino, percent	⚠ 9.1%
Population Characteristics	
i Veterans, 2013-2017	27,604
i Foreign born persons, percent, 2013-2017	35.3%

The Bronx, NYC



i Language other than English spoken at home, percent of persons age 5 years+, 2013-2017	59.3%
Computer and Internet Use	
i Households with a computer, percent, 2013-2017	82.4%
i Households with a broadband Internet subscription, percent, 2013-2017	70.4%
Education	
i High school graduate or higher, percent of persons age 25 years+, 2013-2017	71.5%
i Bachelor's degree or higher, percent of persons age 25 years+, 2013-2017	19.4%
Health	
i With a disability, under age 65 years, percent, 2013-2017	10.9%
i Persons without health insurance, under age 65 years, percent	⚠ 8.9%



US Census Bureau - <https://www.census.gov/quickfacts>



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Income & Poverty	
i Median household income (in 2017 dollars), 2013-2017	\$36,593
i Per capita income in past 12 months (in 2017 dollars), 2013-2017	\$19,721
i Persons in poverty, percent	⚠ 27.9%



Consumer Genomics

- Direct to consumer
- Patient driven



Prenatal



Cancer

Prenatal Genetics – Clinical Guidelines

- Aneuploidy Screening & Diagnosis
 - Cell free DNA
 - First trimester screening
 - CVS and Amniocentesis
- Carrier Screening
 - Hemoglobinopathies
 - Cystic Fibrosis
 - Spinal Muscular Atrophy
 - Fragile X Syndrome

Prenatal Genomics – Clinically Available

- Detection of Microdeletion Syndromes
- Expanded Carrier Screening

Prenatal Genomics – Consumer Driven

Gender Reveal



Teddi Mellencamp

Prenatal Genomics – Consumer Driven

Gender Reveal



[View More on Instagram](#)



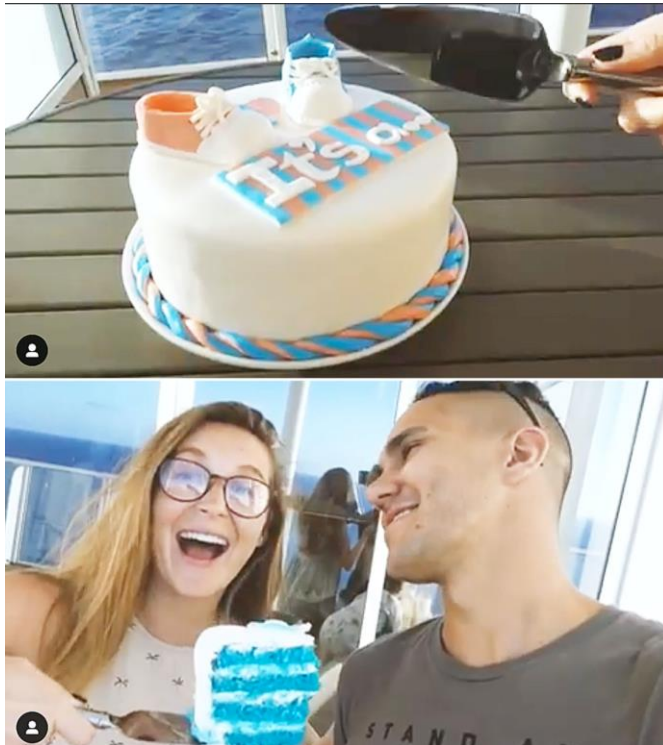
74,113 likes

teddimellencamp

It's a girl! Of course, we would have been equally excited either way, as @tedwinator, the kids and I are just blessed to have a healthy baby no matter the gender. Although I did promise Cruz he could help name her so not sure she'll be too thrilled being called "Catboy" Arroyave. 🙄 Thank you to @ocballoonbar for the pop we needed, @204events for the rad background and @jorgebphoto for capturing this moment so perfectly. 💕

[view all 1,186 comments](#)

Prenatal Genomics – Consumer Driven



Alexa and Carlos PenaVega

"Mommy's outnumbered, but can I say that I love it?" the *Spy Kids* star said in a March 2019 video after revealing the sex of her baby with a colorful cake. "The idea of two boys is so exciting. It's such a blessing. I have a beautiful momma's boy already, and now I get two!"

Prenatal Genomics – Consumer Driven

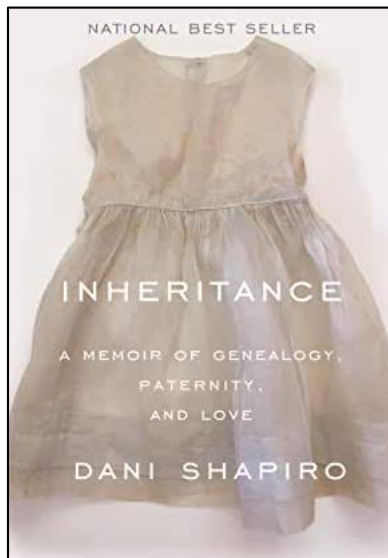


Prenatal Genomics – Gender Reveal

- Patients want early information and are often under pressure ... because the party is already planned.
- Patients want the results to be given to someone else – sister, bakery, etc.
- Patients frequently embark on the “gender test” with little consideration that the aneuploidy screening test results might be high risk.
- The patient interface – My Chart – does not display “send out” labs and so patients often misinterpret the Fragile X screening result as the gender.

Prenatal Genomics – Paternity Testing

- Many mothers are interested – prospectively
- Home DNA kits are advertised for holiday gifts and stocking stuffers:
 - Blogs and current books discuss family issues raised by unexpected results



Cancer Genomics – Consumer Driven

- How to get the right patients to Genetics?
 - Often see the worried well
 - Some individuals from high risk families are never seen in Genetics
- Patients are quite interested in multi-gene panels
 - May find something unexpected

Cancer Genomics – Consumer Driven

- Variants of Uncertain significance
 - Surgeons might plan to operate based on a VUS
- What to do with a VUS?
 - Patients often report that they will eat a healthier diet and exercise

Cancer Genomics – Consumer Driven

- When it is believed that there is something in the family ...
- It's hard to get copies of previous testing results from family members
- Patients or family members can't always remember the results from testing they undertook previously
- There can be confusion as results are conveyed from one family member to another
 - PALB2 vs BRCA2

Cancer Genomics – Consumer Driven

- Lab reports are scanned into EPIC as “send out” lab reports and are not easily accessible to other providers or to patients via MyChart

The Current Landscape of Consumer Genomics

- Clinical Utility
 - Planning a party
 - Evaluating an ultrasound anomaly
 - Eating healthy
- Communicating and keeping track of results
 - Reports are hard to understand & keep track of
 - Sharing information within families
 - EMRs and interoperability



Thank you!