

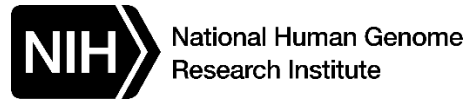
C G T A C G T A
A C G T A C G T

Welcome from NHGRI

Ben Solomon, MD

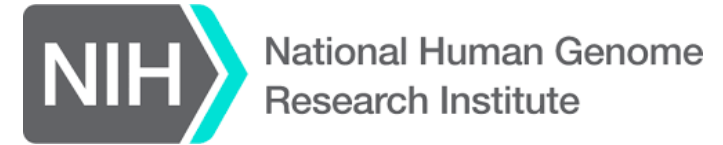
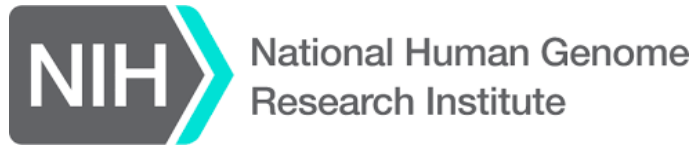
Clinical Director, NHGRI

April 5, 2024



A little about who I am and how I got here...

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The main thing I have learned!

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A C G





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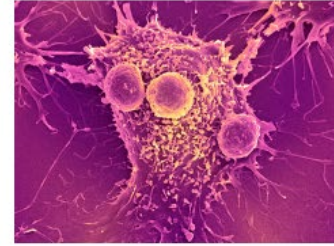
news & views

CLINICAL GENETICS

Time to make rare disease diagnosis accessible to all

Studies have demonstrated the value of genomic analysis for the diagnosis of rare diseases, but accessibility is still in its infancy; global data sharing is needed to further advance our knowledge of all causes of rare disease.

NHGRI IRP



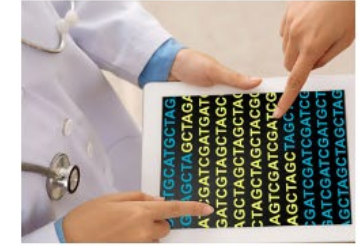
Cancer Genetics and Comparative Genomics Branch >

The NHGRI Cancer Genetics and Comparative Genomics Branch identifies and study genes that contribute to cancer susceptibility and progression.



Center for Genomics and Data Science Research >

NHGRI center focused on computationally intensive approaches to analyze large-scale genomic data and identifying genetic contributions to human disease.



Center for Precision Health Research >

The Center for Precision Health Research, formerly Medical Genomics and Metabolic Genetics Branch, studies how health and disease are related to genetic, genomic and metabolic variation.



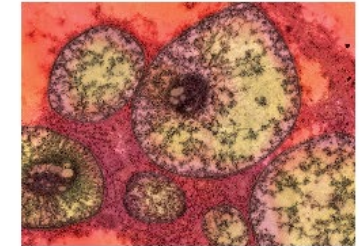
Center for Research on Genomics and Global Health (CRGGH) >

CRGGH facilitates understanding of the relationship between genetic variation and population differences in disease distribution to inform health disparities.



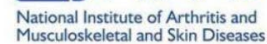
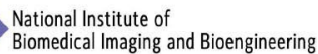
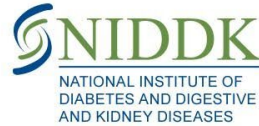
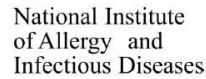
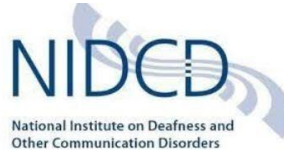
Medical Genetics Branch >

The Medical Genetics Branch studies inherited disorders of metabolism and of human development.



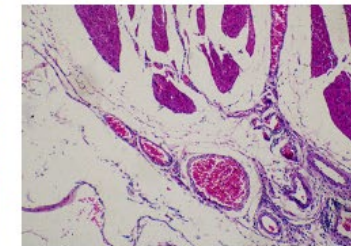
Metabolic Medicine Branch >

The Metabolic Medicine Branch houses clinical and laboratory research programs that focus on the development of new therapies for patients with genetic disorders.



Social and Behavioral Research Branch >

The Social and Behavioral Research Branch conducts research to apply genomic discoveries to improve health and clinical care.



Translational and Functional Genomics Branch >

The Translational and Functional Genomics Branch explores the genetic, epigenetic and metagenomic basis of human disorders.

NHGRI clinical studies

- 80 non-exempt human subjects research studies
 - 67 observational studies
 - 13 interventional protocols (from psychosocial designs to gene therapy trials)



Three examples of things I am particularly excited about...

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1. New therapies

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LOCAL

Their three children have a fatal disease, and they refused to let the coronavirus take away their only chance at saving them



Perspective by [Theresa Varvas](#)
Metro columnist

April 25, 2020 at 9:00 a.m. EDT



Jessica Lindqvist and Niclas Flysjo at their home in Sweden with their children, left to right, Hampus, Julia and Isabella. (Courtesy of Niclas Flysjo)



Learn more about Dr. Venditti's research on methylmalonic acidemia (MMA) in this video.



Proteus Syndrome Treatment - Leslie Biesecker with patient Jerry DeVries

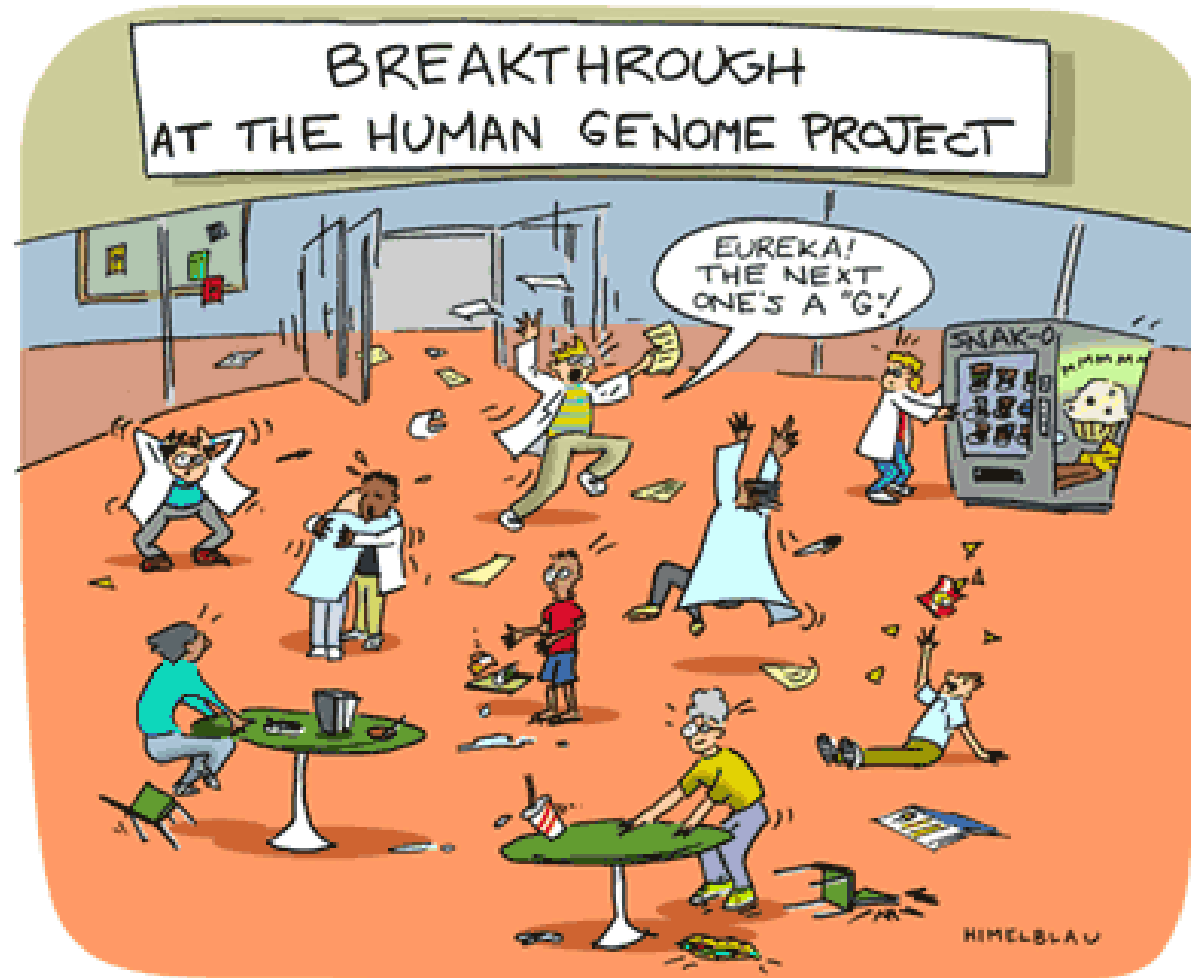
National Human Genome Research Institute
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2. Figuring out ways to get more answers

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A C G



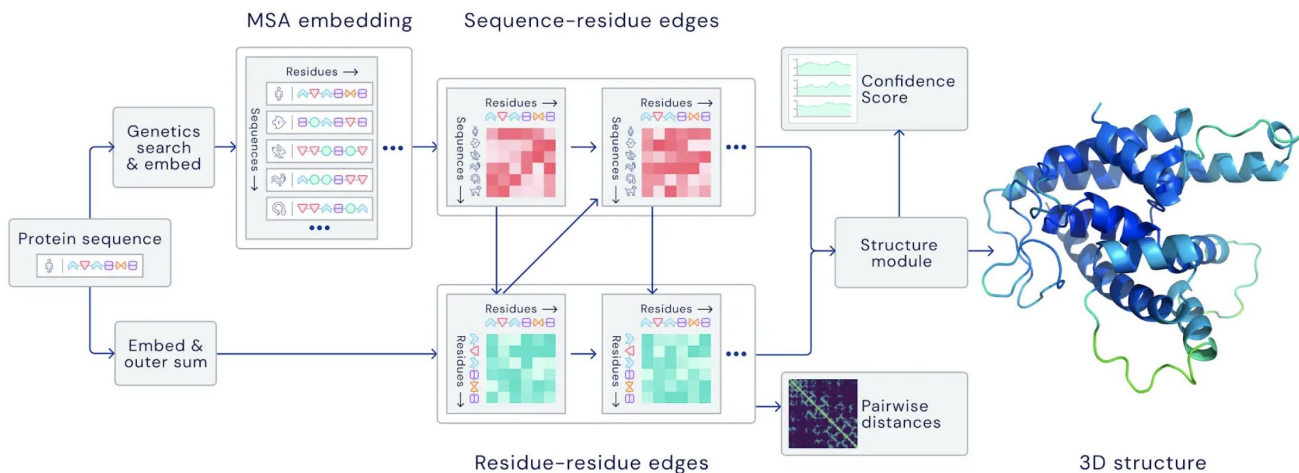


genome.gov; Time magazine

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3. AI (of course!)

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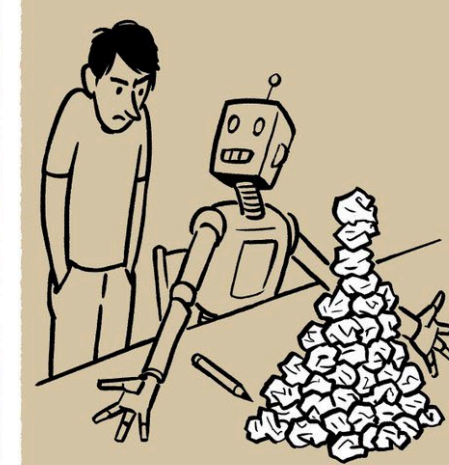
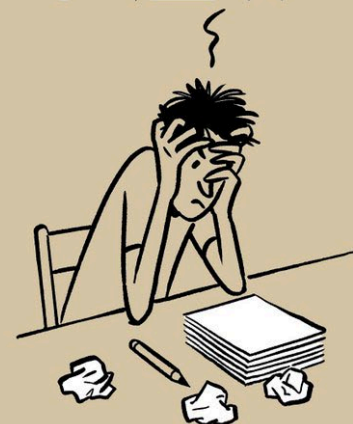


PRICE \$8.99

NOV. 20, 2023

THE NEW YORKER

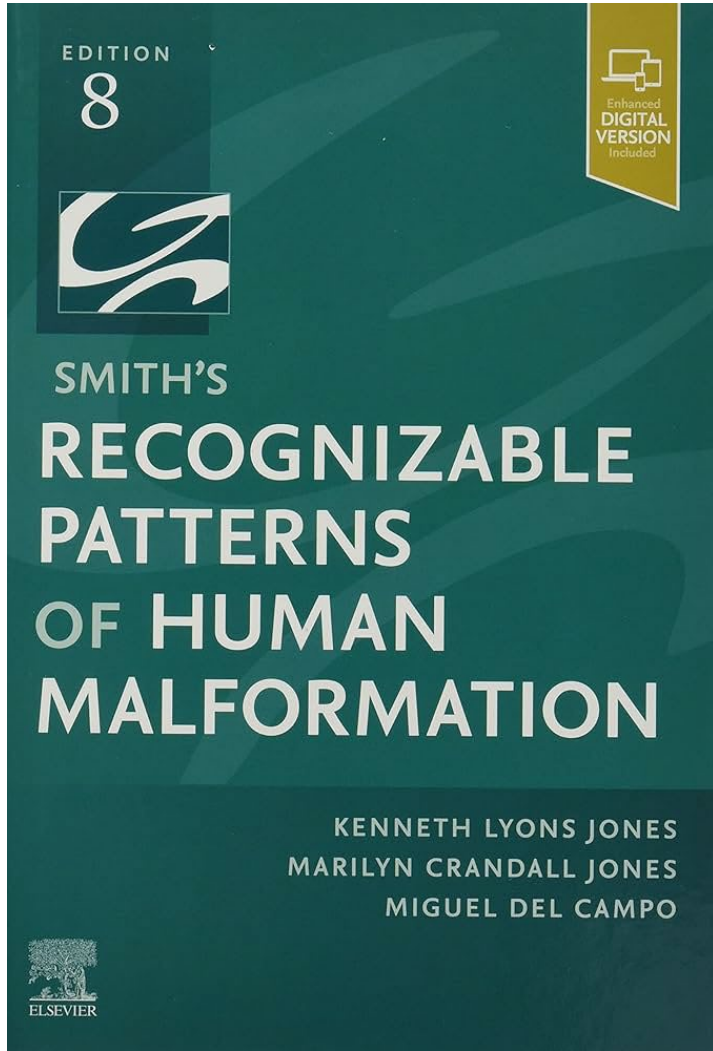
CREATE YOUR OWN COVER



Nature's 10
 Ten people (and one non-human) who helped shape science in
 2023
 13 DECEMBER 2023

Image: Kim Albrecht; artificial-worldviews.kimalbrecht.com





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Noise-thresholds on visual heat maps

None

Low

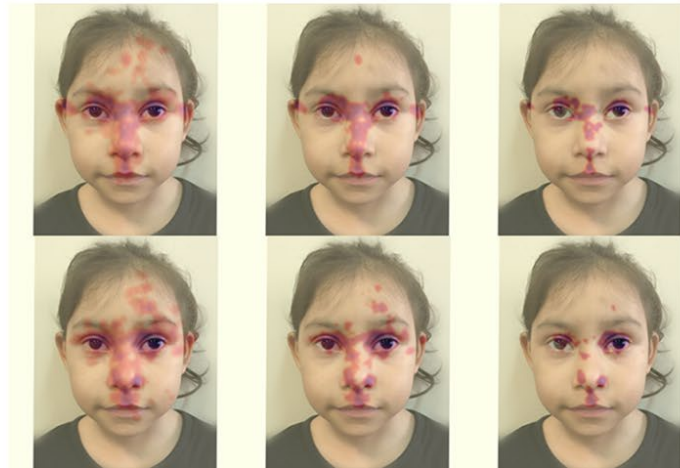
High

Coverage-thresholds applied to model saliency maps

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Successful clinicians

Successful nonclinicians



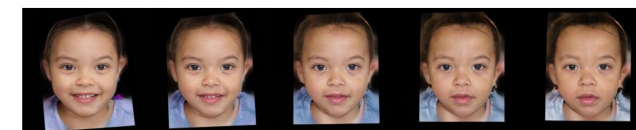
Unaffected → Noonan Syndrome



Unaffected → Noonan Syndrome

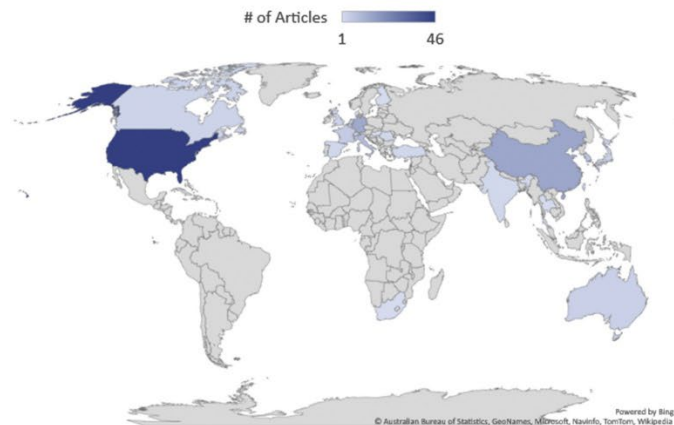


Unaffected → Noonan Syndrome

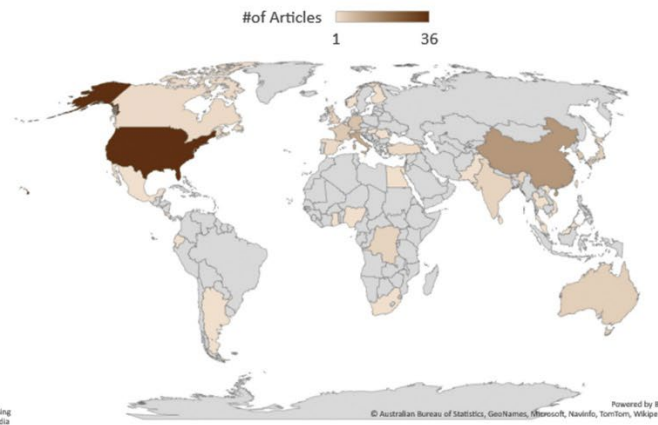


Unaffected → Noonan Syndrome

A Country of Corresponding Author(s) (n = 141 corresponding authors from 134 articles)

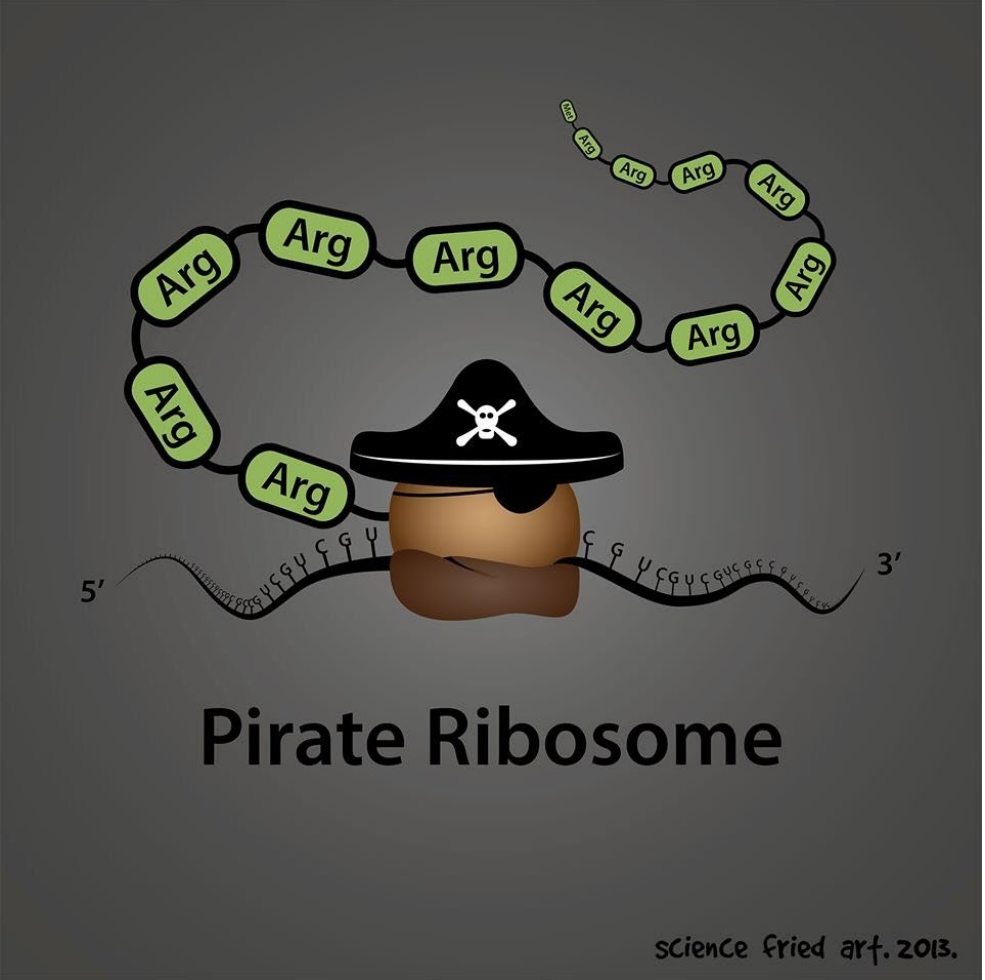
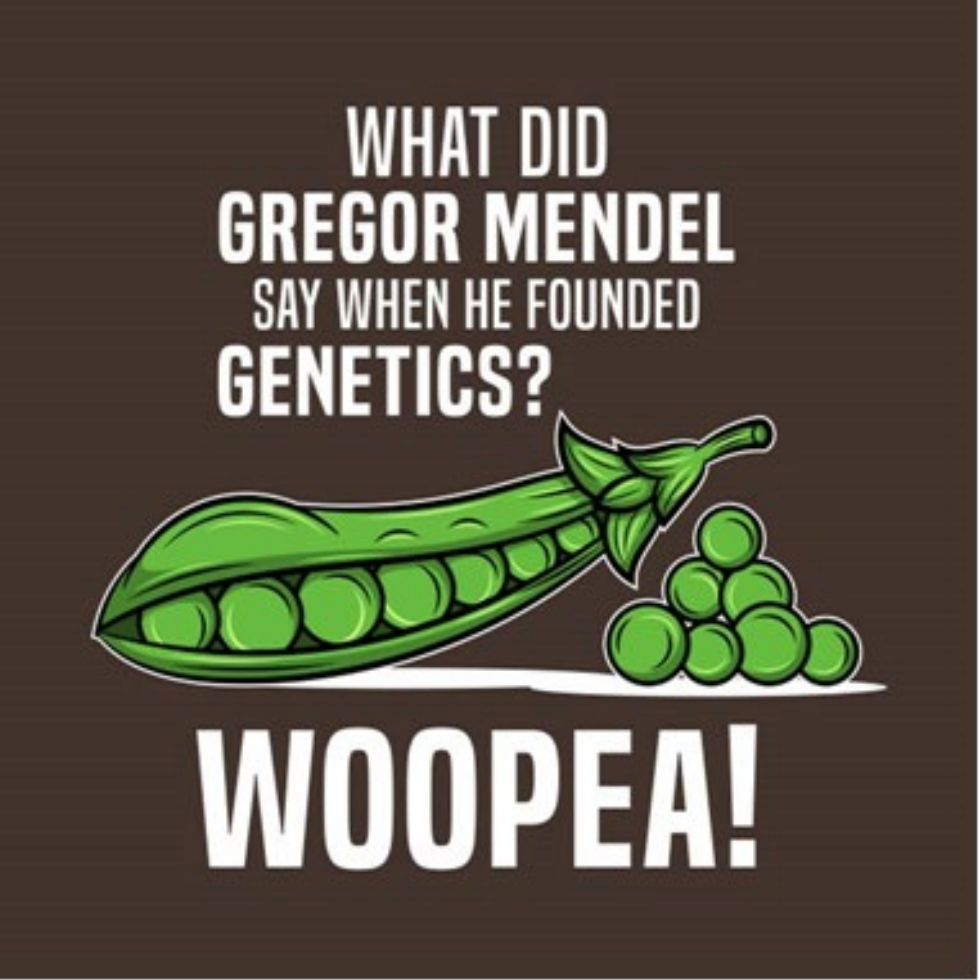


B Country of Studied Populations (where specified) (n = 167 populations from 119 articles where country-specific were available)



Thank you!

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A C G





—
The **Forefront**
of **Genomics**[®]
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