PERSONAL GENOME PROFILE 1:

Rosalynn Gill was one of the first ten Personal Genome Project participants (PGP9). Hers was the first fully public genome of a human female. Each of these participants have shared their genomic and medical information with the world

Where do I find her data?

Rosalynn's information is on the personalgenomes.org site as **hu034DB1**. Her public profile can be found at https://my.pgp-hms.org/profile/hu034DB1

Background?

Rosalynn was 45 years old in 2007 when she had her genome sequenced. She had to consider how her decision to be sequenced and release the information to the world would affect her children. She had some concerns about a family history of cancer and cardiac disease, but wanted all of the information.

A video about her background and her approach to this project an be found in

GENOME: The Future is Now WEBISODE 2 (https://www.youtube.com/watch?v=2r9DpthvNKM)

and further interviews are available in the personalgenomes.org youtube channel and playlists.

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Variant	Clinical Importance	Impact	Allele freq	Summary
APOE-C130R	High	Well-established pathogenic Complex/Other, Heterozygous	14%	This is generally known as the ApoE4 allele of ApoE and is associated with increased risk of Alzheimer's. 20-25% of individuals are heterozygous for this variant, and 1-2% are homozygous. Data from Khachaturian et al. suggests an average 7% of all individuals developed Alzheimer's by the age of 80; when this is split by ApoE4 status: 10% of ApoE4 heterozygotes (3% increased attributable risk), 40% of ApoE4 homozygotes (33% increased attributable risk), and 5% of non-carriers (2% decreased attributable risk). Notably, their model suggests 70-75% of people would eventually develop Alzheimer's by the age of 100 regardless of ApoE4 genotype (and 25-30% are resistant, regardless of genotype), but that ApoE4 variants shift the disease onset to occur significantly earlier (4 years earlier for heterozygous carriers, 13 years for homozygotes).
SCN5A-G615E	High	Uncertain pathogenic Dominant, Heterozygous	0.029%	Rare, reported to be associated with long-QT syndrome (can cause syncopal spells, sudden death as a teenager / young adult), but observations are scattered may have some publication bias.
MTR-P749S	High	Uncertain pathogenic Unknown, Heterozygous	0.10%	Unreported, predicted to be damaging. Other recessive missense mutations in this gene cause methylcobalamin deficiency.

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