

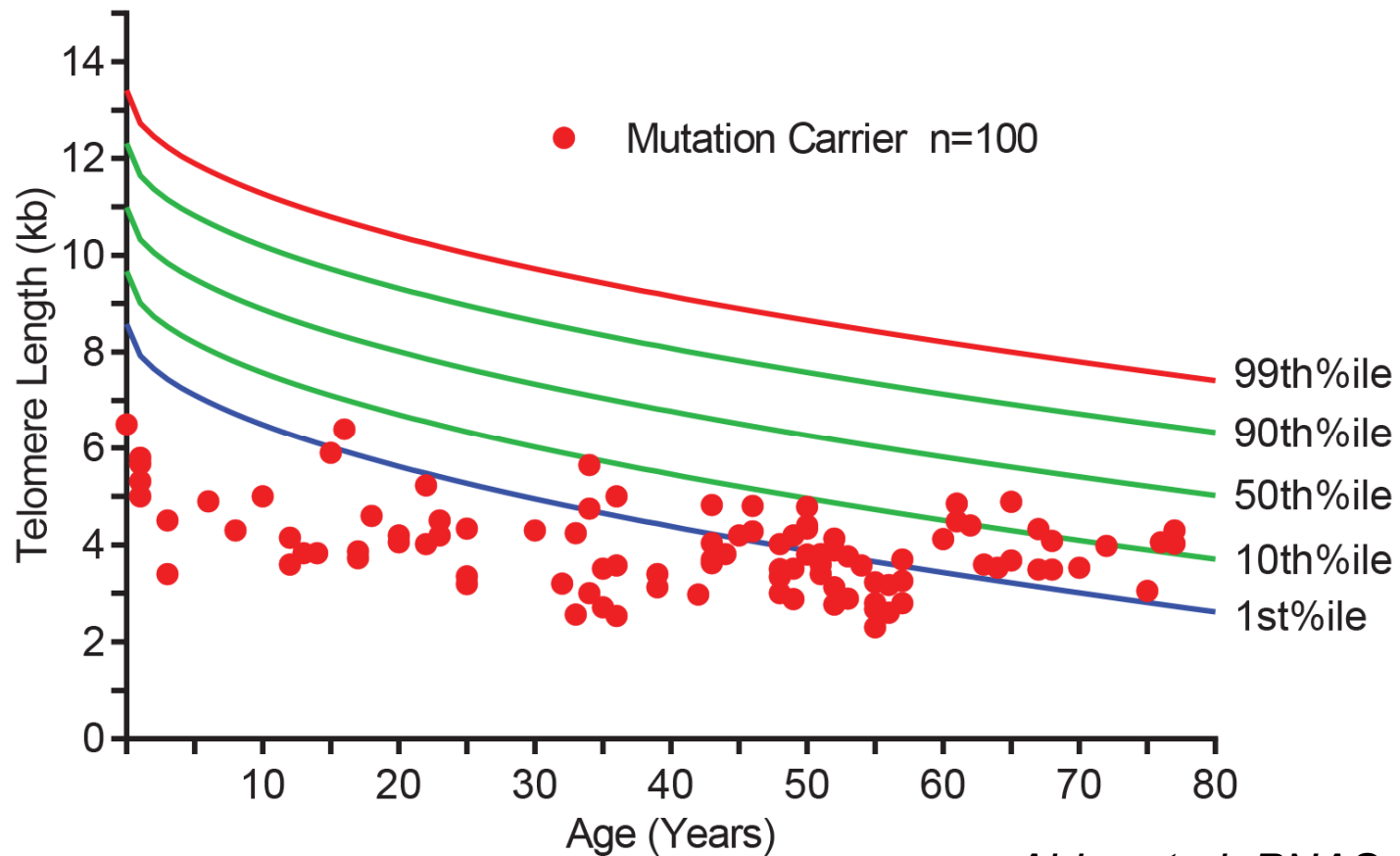
The \$99 Question: Can a DIY Home Test Really Tell Your Biological Age?

Mary Armanios

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Science Writers' Boot Camp-DC
May 7, 2018

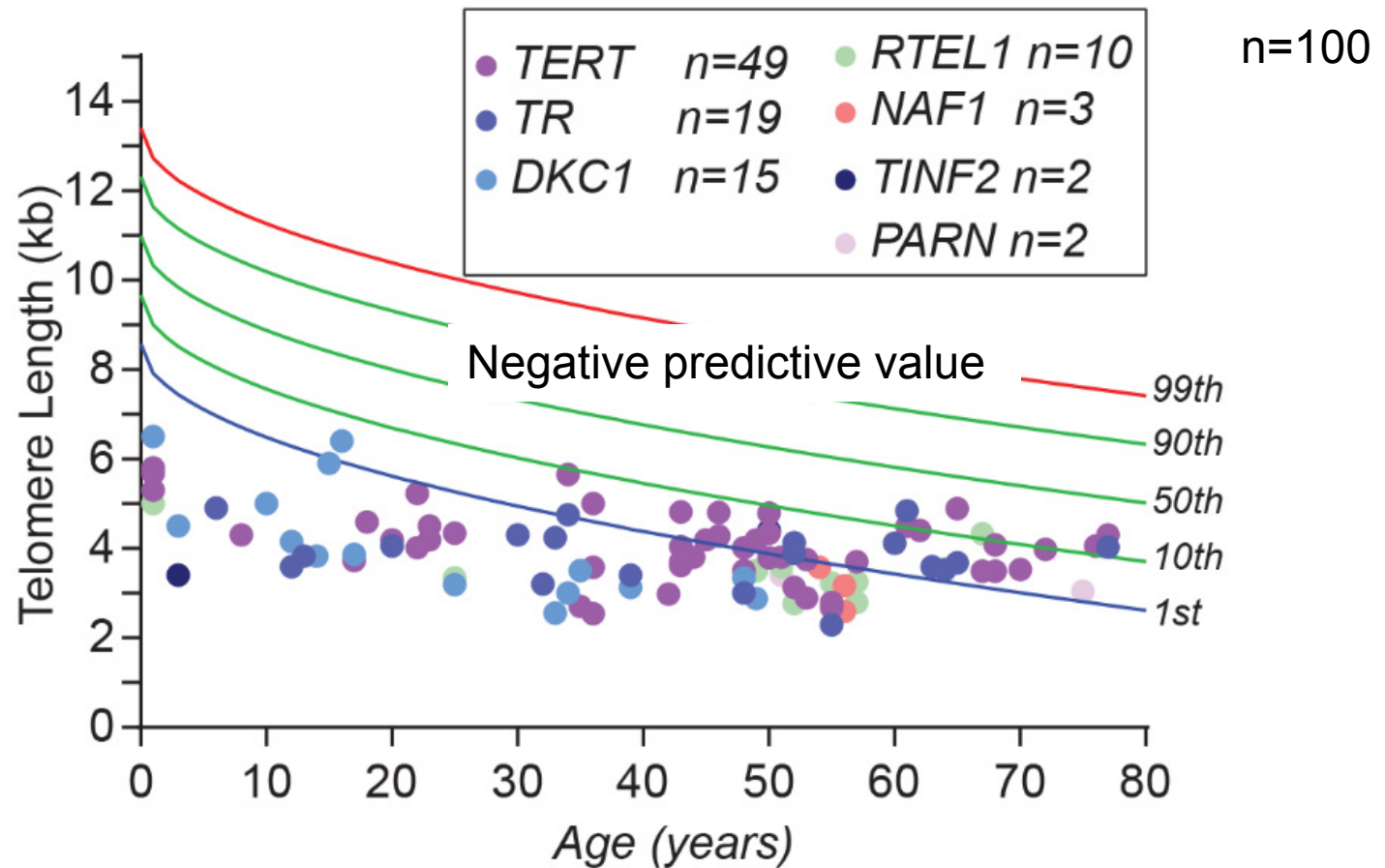
Abnormally short telomere length causes disease



Alder et al. PNAS 2018

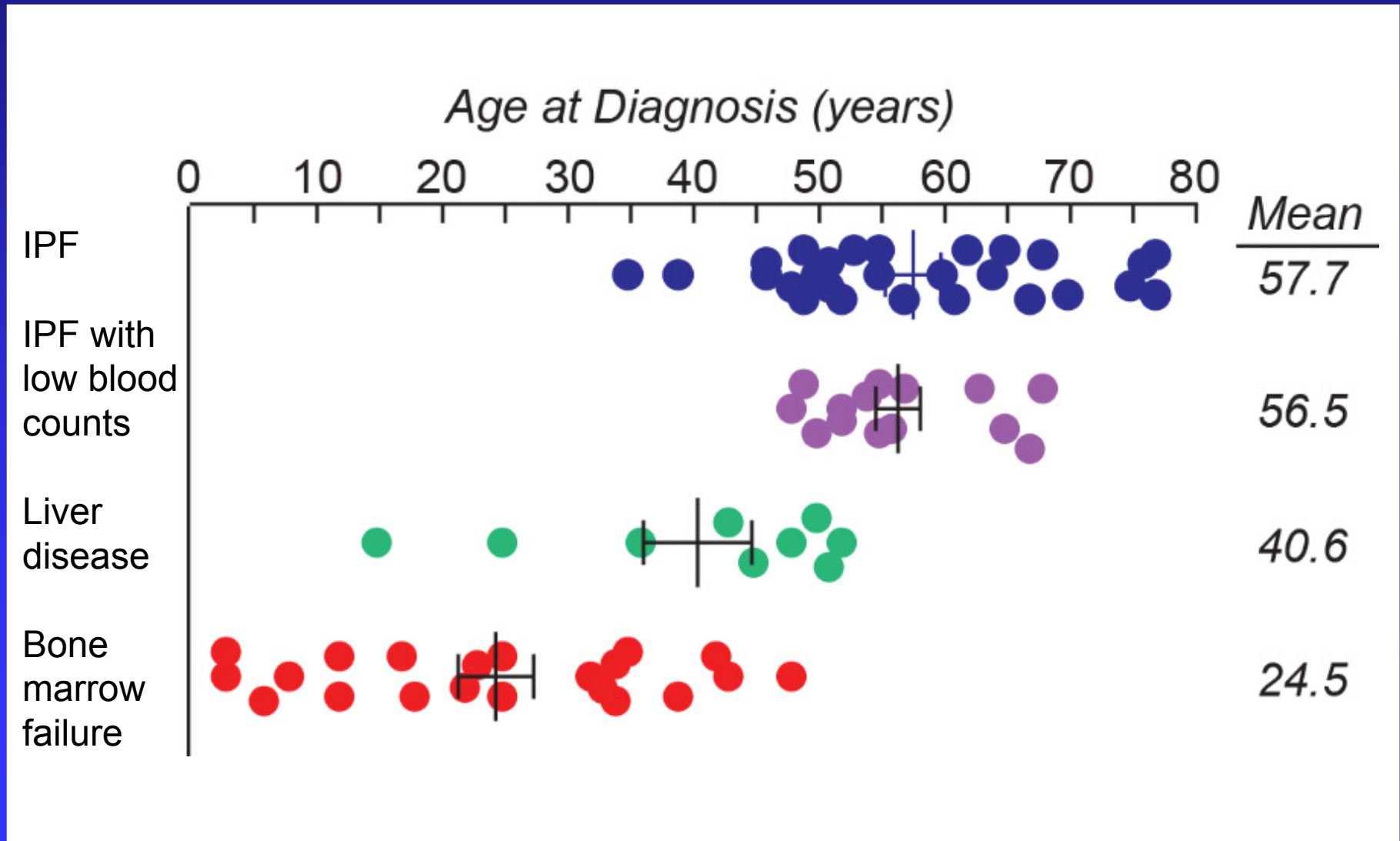
Clinically available test, Johns Hopkins Pathology Labs

Telomerase mutations carriers have short telomeres

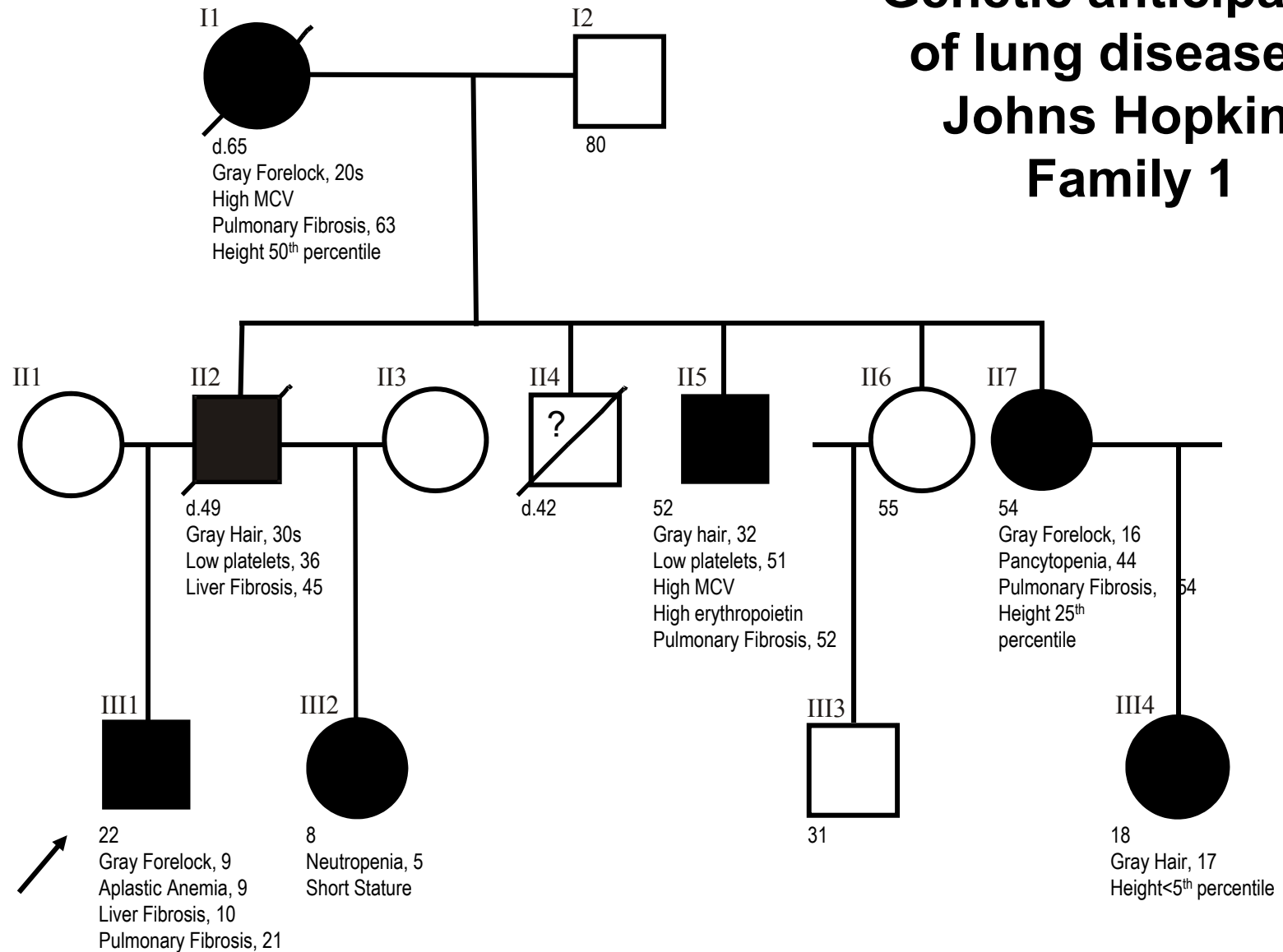


Alder et al.

The manifestations of telomere-mediated disease are age-dependent



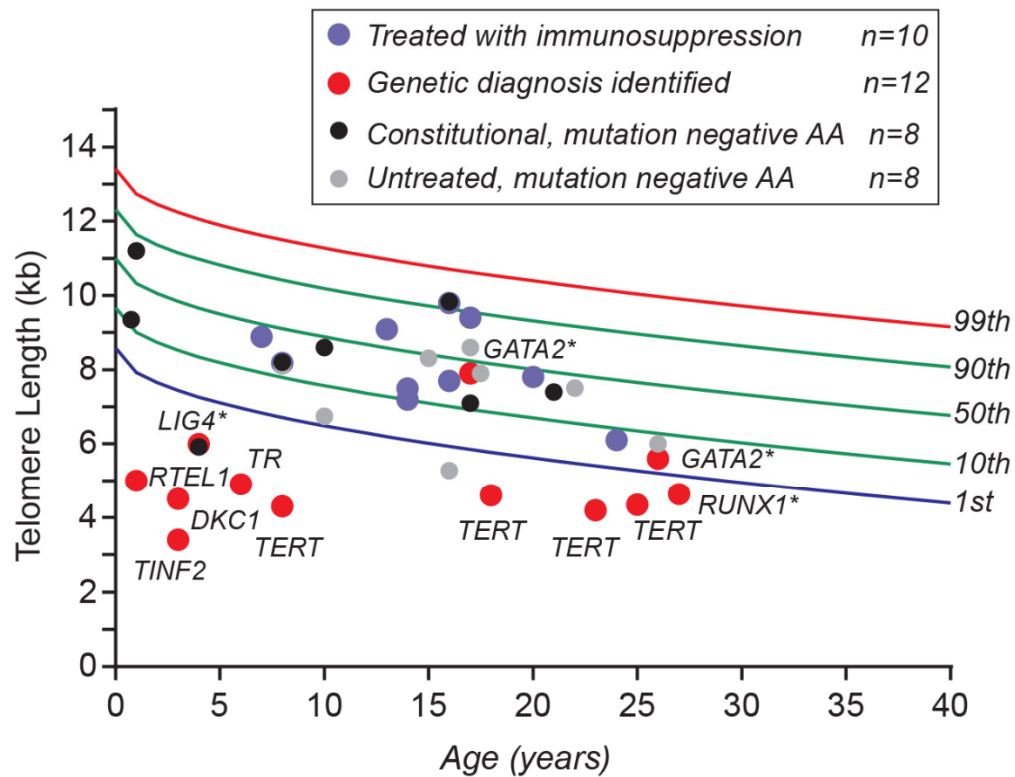
Genetic anticipation of lung disease in Johns Hopkins Family 1



TERT K902N

Armanios *PNAS* 2005

Telomere length results altered a major clinical decision in ~25% of cases





Diagnostic utility of telomere length testing in a hospital-based setting

Jonathan K. Alder^{a,b,1}, Vidya Sagar Hanumanthu^{a,b}, Margaret A. Strong^{b,c}, Amy E. DeZern^{a,d}, Susan E. Stanley^{a,b}, Clifford M. Takemoto^e, Ludmilla Danilova^a, Carolyn D. Applegate^{a,b,f}, Stephen G. Bolton^g, David W. Mohr^f, Robert A. Brodsky^{a,d}, James F. Casella^e, Carol W. Greider^{a,b,c,2}, J. Brooks Jackson^g, and Mary Armanios^{a,b,c,f,g,2}

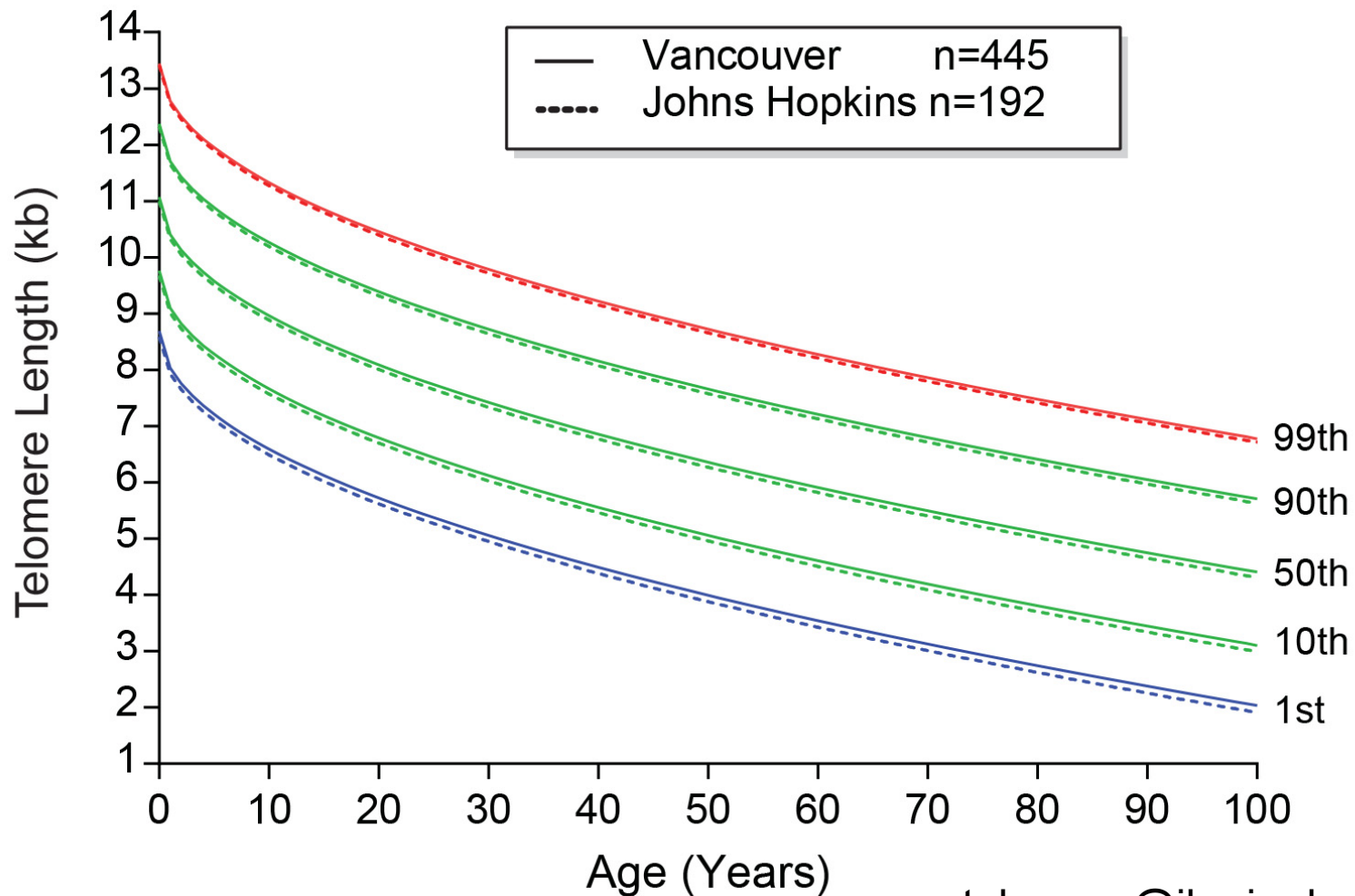
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^bTelomere Center at Johns Hopkins, The Johns Hopkins University School of Medicine, Baltimore, MD 21287; ^cDepartment of Molecular Biology and Genetics, The Johns Hopkins University School of Medicine, Baltimore, MD 21287; ^dDepartment of Medicine, The Johns Hopkins University School of Medicine, Baltimore, MD 21287; ^eDepartment of Pediatrics, The Johns Hopkins University School of Medicine, Baltimore, MD 21287; ^fMcKusick-Nathans Institute of Genetic Medicine, The Johns Hopkins University School of Medicine, Baltimore, MD 21287; and ^gDepartment of Pathology, The Johns Hopkins University School of Medicine, Baltimore, MD 21287

Contributed by Carol W. Greider, January 9, 2018 (sent for review November 28, 2017; reviewed by Thomas R. Cech and Agata Smogorzewska)

Feb 20, 2018

Telomere length has discrete, definable boundaries in the human population

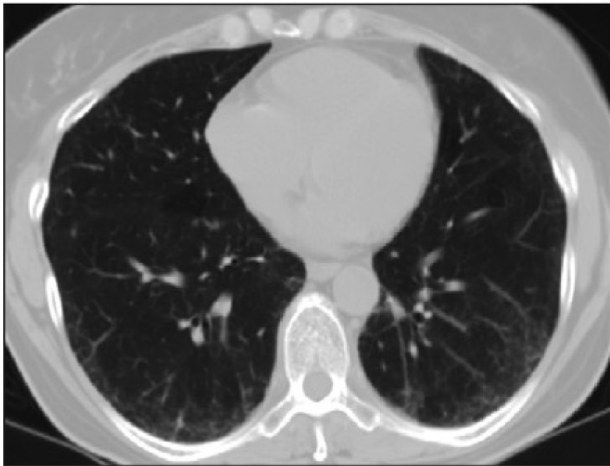


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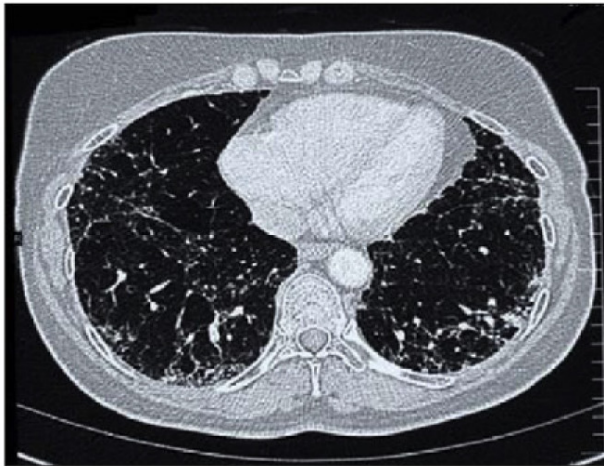
Data from CLIA/CAP certified assay Johns Hopkins Pathology; Alder *et al.* *PNAS* 2018 compared with Aubert *et al.* *PLoS Genetics* 2012

Lung disease is the third leading cause of death in the United States

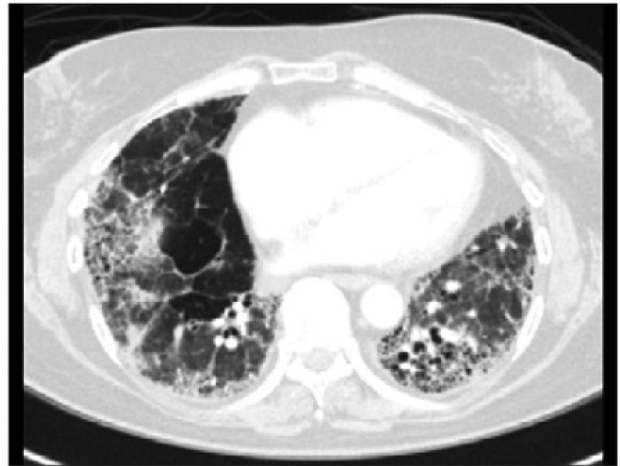
Age 54



Age 59



Age 61

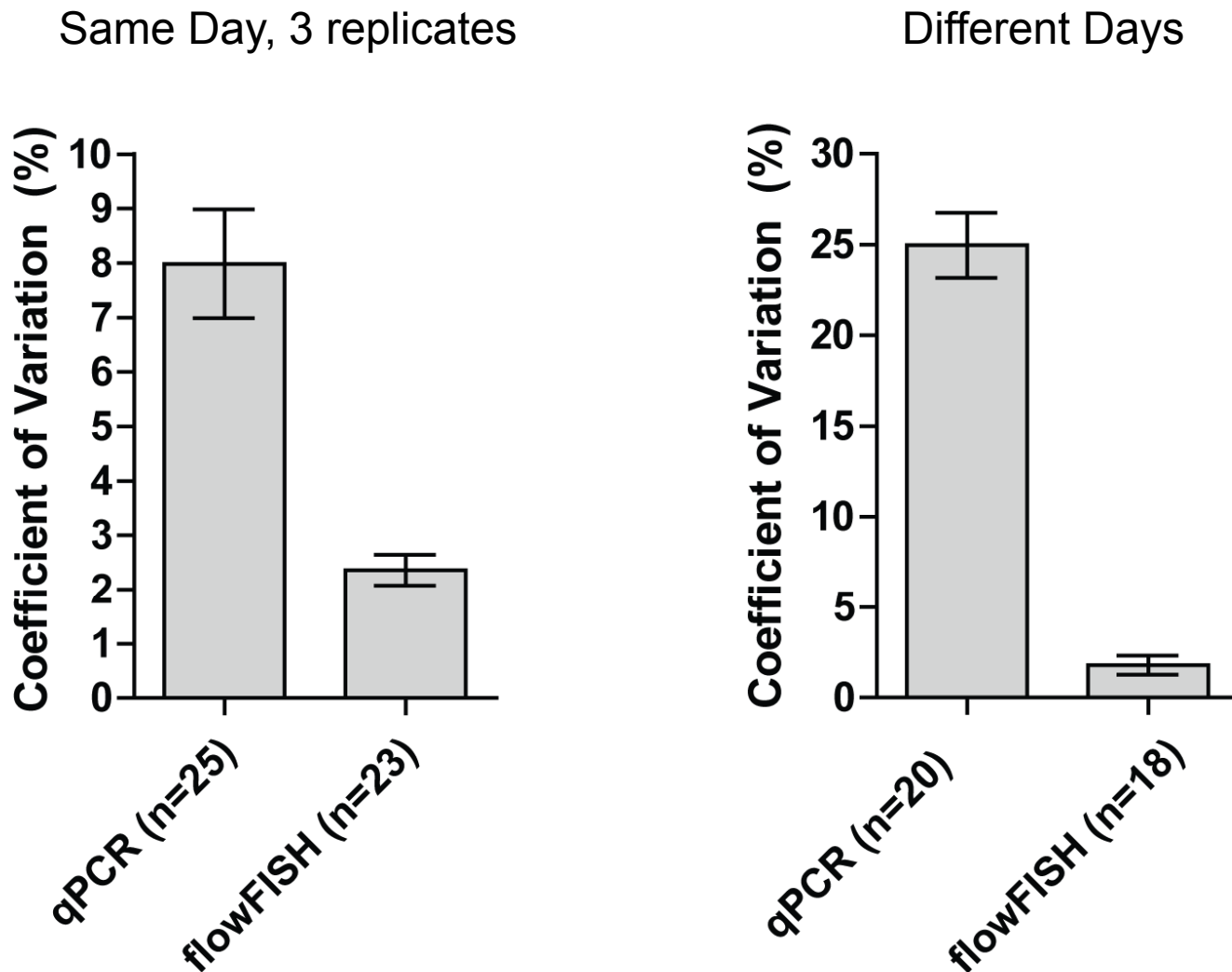


Why a DIY \$99 test cannot tell biological age?

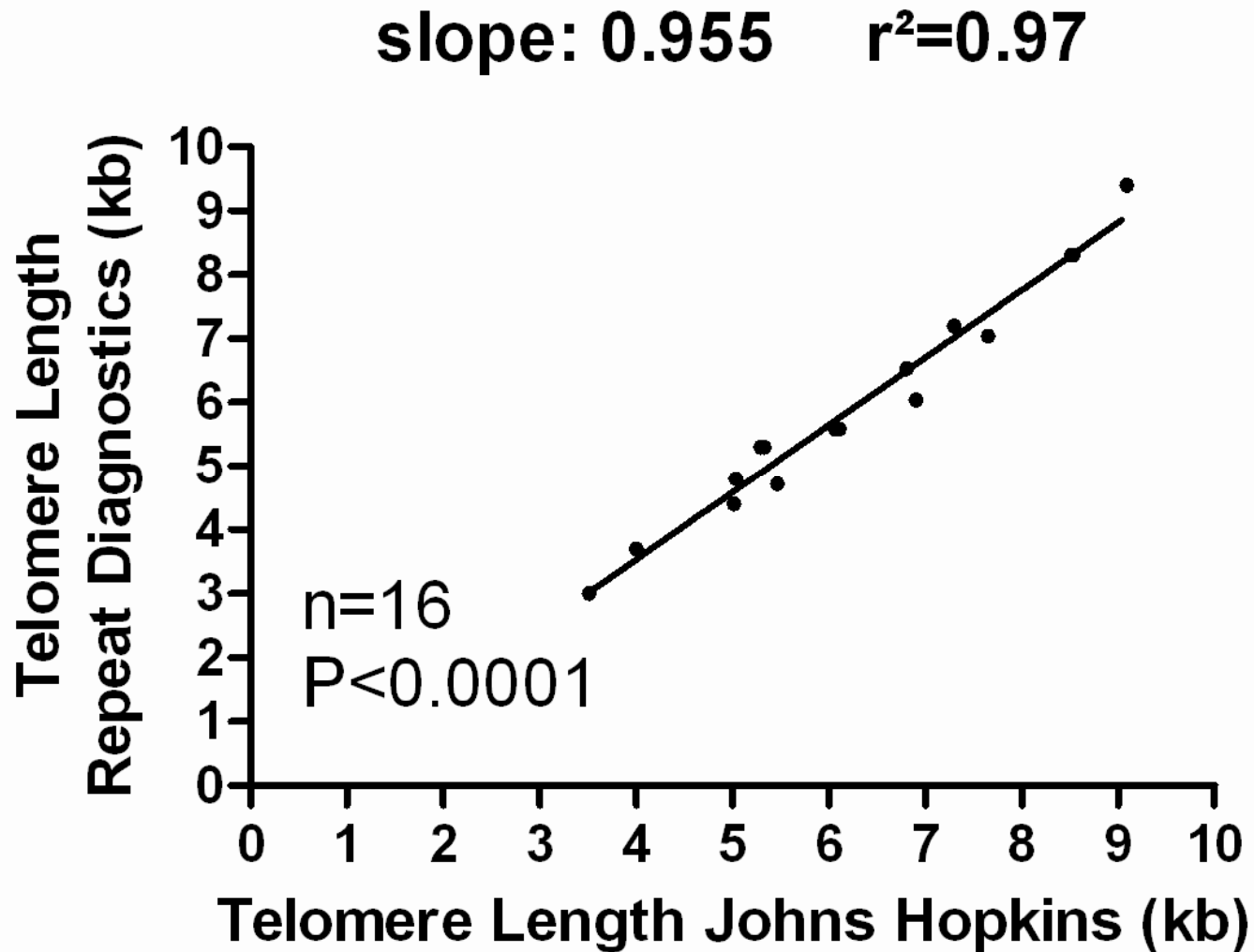
- *Reproducibility arguments*
- *Interpretation issues*
- *Biological arguments*

and potential harm

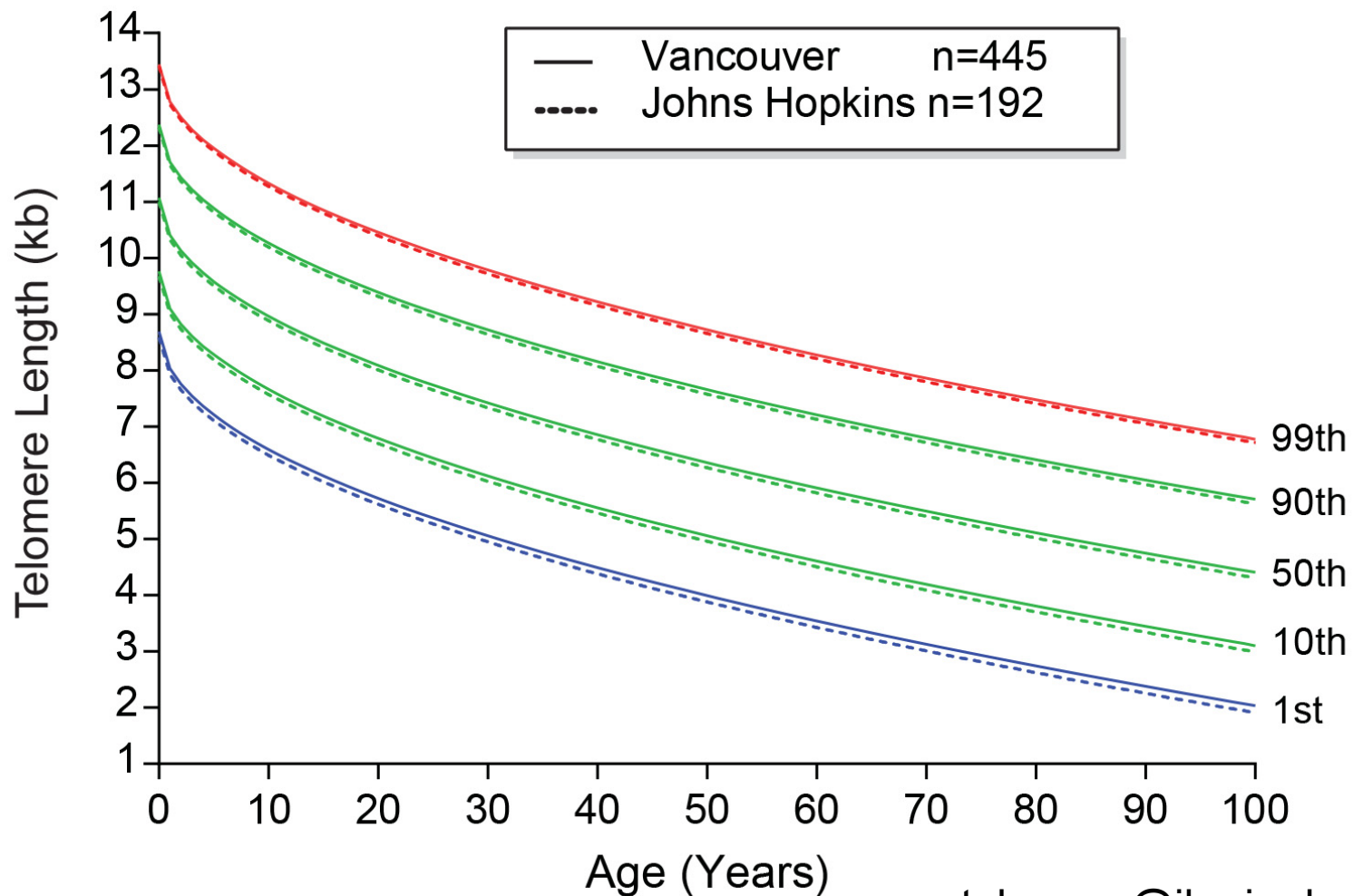
Telomere length measurement by flowFISH is highly reproducible



Outstanding inter-lab correlation



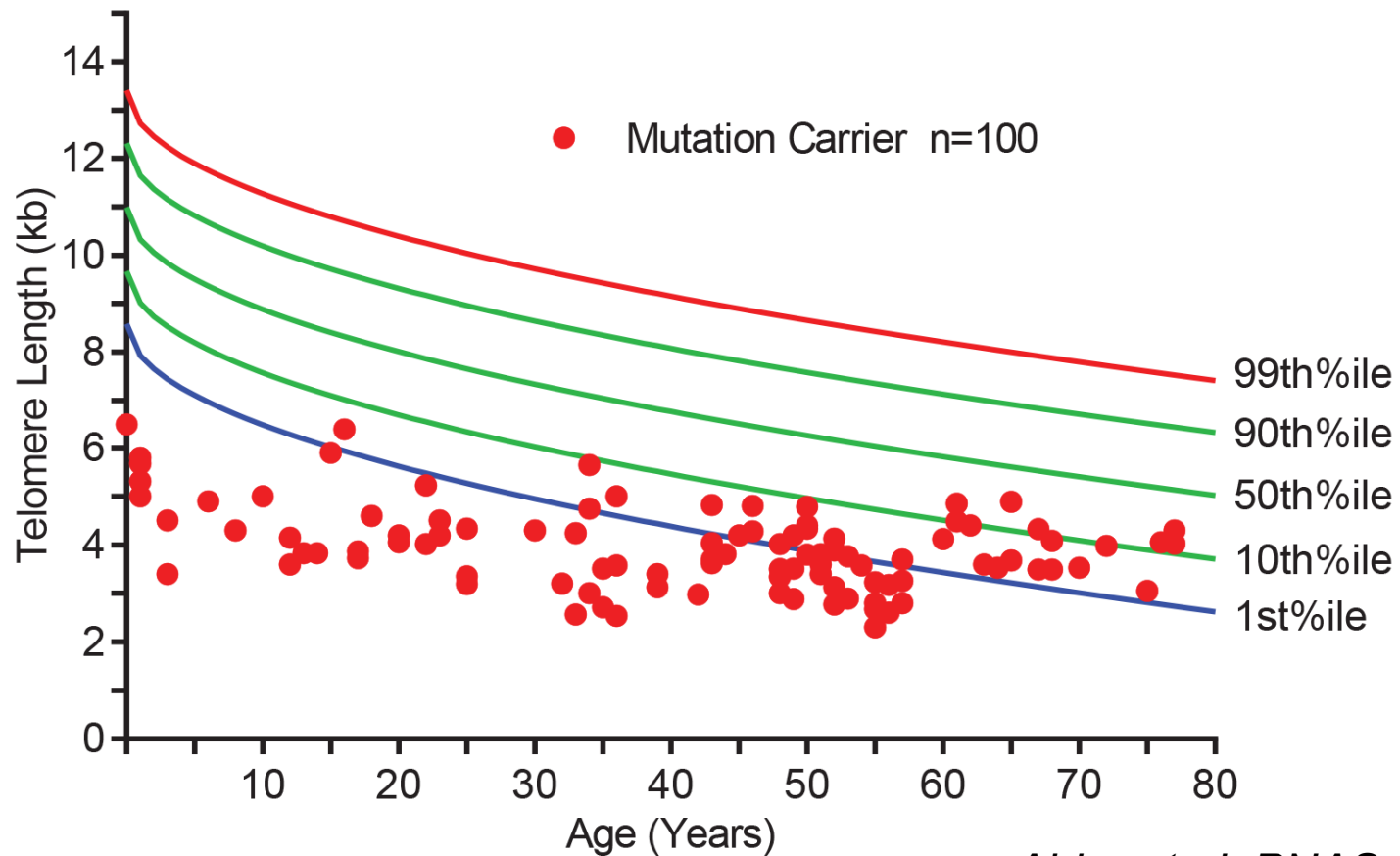
Telomere length has discrete, definable boundaries in the human population



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Data from CLIA/CAP certified assay Johns Hopkins Pathology; Alder *et al.* *PNAS* 2018 compared with Aubert *et al.* *PLoS Genetics* 2012

Telomere length by flow cytometry and FISH is the gold standard for clinical use



Alder et al. PNAS 2018

Clinically available test, Johns Hopkins Pathology Labs

Summary

- Short telomeres cause a group of discrete clinical phenotypes; their recognition is critical for patient care decisions
- The telomere length distribution is definable; harm at both extremes
- Telomere length measurements available to the public use problematic methodologies and provide information that is not evidence-based; argument for potential harm