

11.20.2023 CARS2 REBUTTAL



ARIZONA DEPARTMENT
OF HEALTH SERVICES
ARIZONA BIOMEDICAL RESEARCH CENTRE



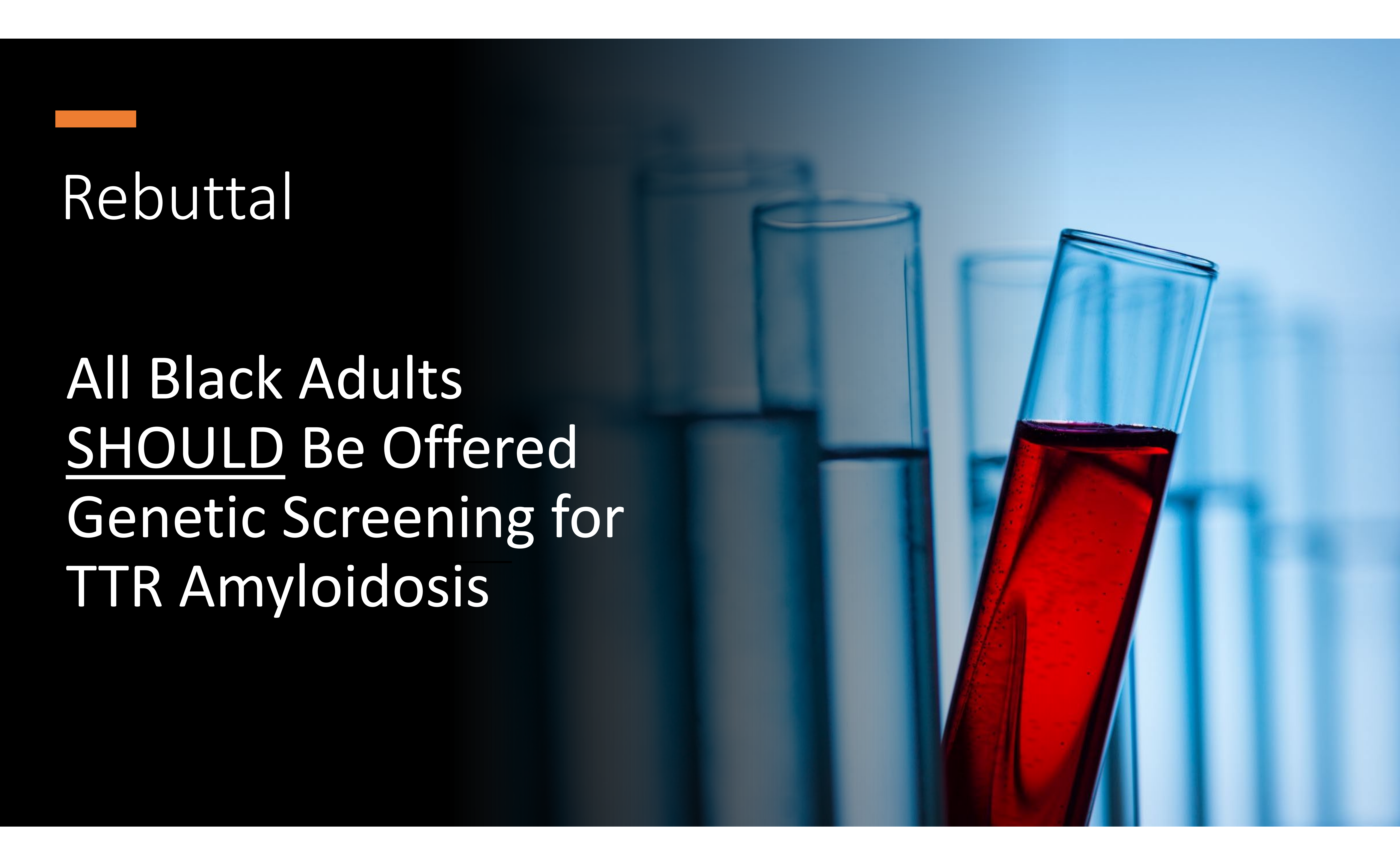
FUTURE
HEALTH





Rebuttal


All Black Adults
SHOULD Be Offered
Genetic Screening for
TTR Amyloidosis





Rebuttal

All Black Adults
Should NOT Undergo
Genetic Screening for
TTR Variants



Rebuttal...Well, maybe....

CardioNext®: Analyses of 92 Genes Associated with Inherited Cardiomyopathies and Arrhythmias

RESULTS

TTR

Pathogenic Mutation: p.V142I

SUMMARY

POSITIVE: Pathogenic Mutation Detected

INTERPRETATION

- This individual is heterozygous for the p.V142I (c.424G>A) pathogenic mutation in the *TTR* gene.

Rebuttal...Well, maybe....

- Background: (pV142I)

A 76-year-old male (Black) with past medical history including **systolic and diastolic HF-**

- The patient has had **multiple emergency department visits with shortness of breath and thoracenteses.**
- **Orthopnea and sleeping in a chair** since discharged from the hospital in April 2023.
- **ECGs show low voltage in all limb leads.**
- **Bilat carpal tunnel with repairs.**
- **Echo thick walls with thickening of MV and AV leaflets.**
- **Recurrent Pleural effusions**
- **Family History:**
 - **He had 6 siblings who all had carpal tunnel, and some were diagnosed with “fluid buildup”. His youngest sister died last year from “fluid in her lungs”.**
 - **Multiple older relatives with “dropsy” but they never knew what caused it. (several generations).**
 - **He has 5 children**
 - **3 sons**
 - **2 daughters. Oldest daughter is 41 years old and already had carpal tunnel surgery. (After patients’ diagnosis she had genetic testing and has the same genetic abnormality.)**



Tab Blackshear



Questions?

A stage with red sequined curtains and a gold fringe valance. The curtains are heavily textured with small, reflective sequins that catch the light, creating a shimmering effect. The valance at the top is made of a gold, shimmering fabric with a fringe edge. The overall scene is warm and celebratory.

Final Remarks