Clinical Care Implementation of Guidelines for Genomic Testing

The impact of disparities and discrimination on implementation:

Lessons learned from sickle cell genetic screening

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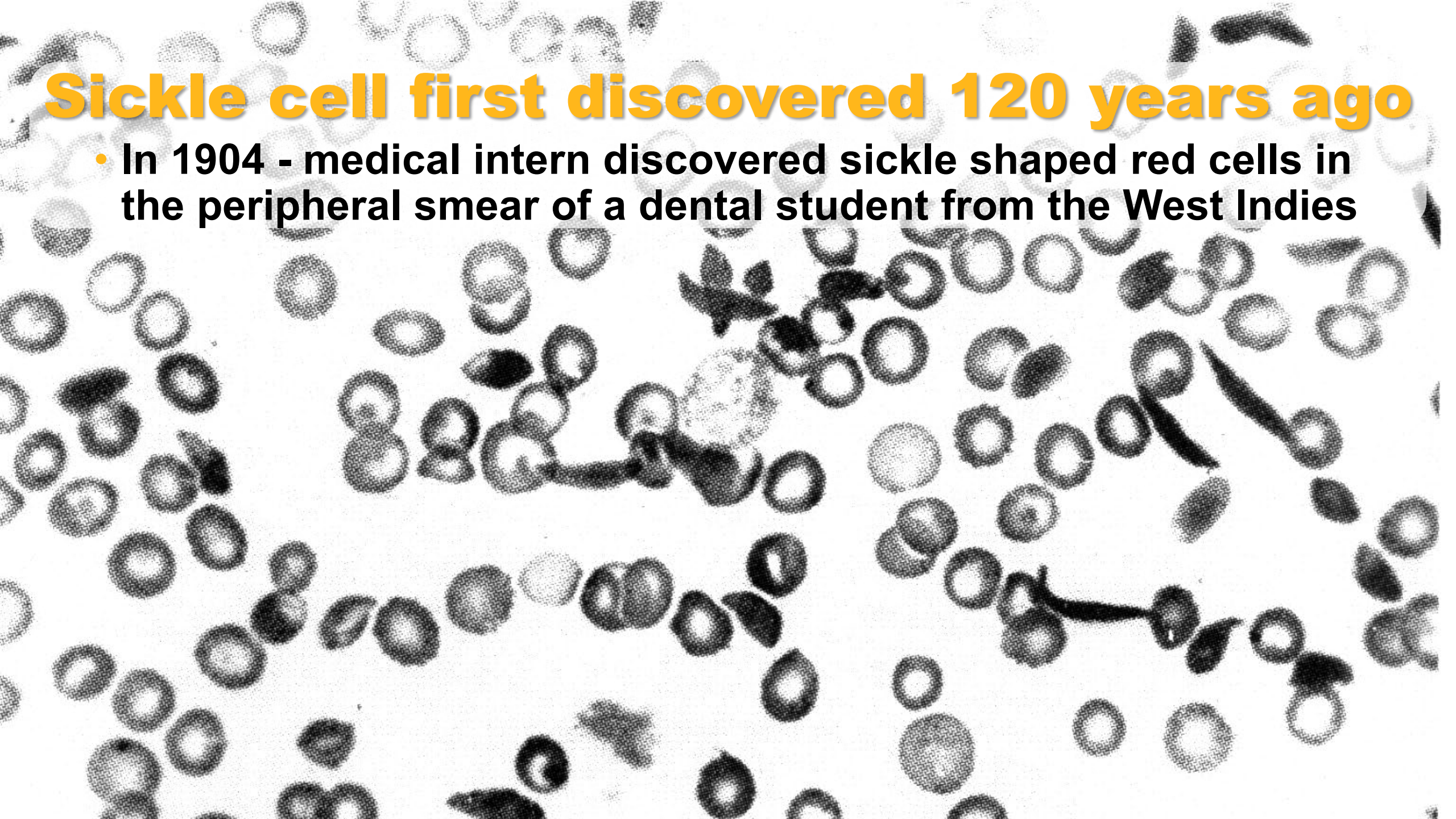
Financial disclosure

Dr. Jonassaint is the founder and owner of Expressive Painimation Co.

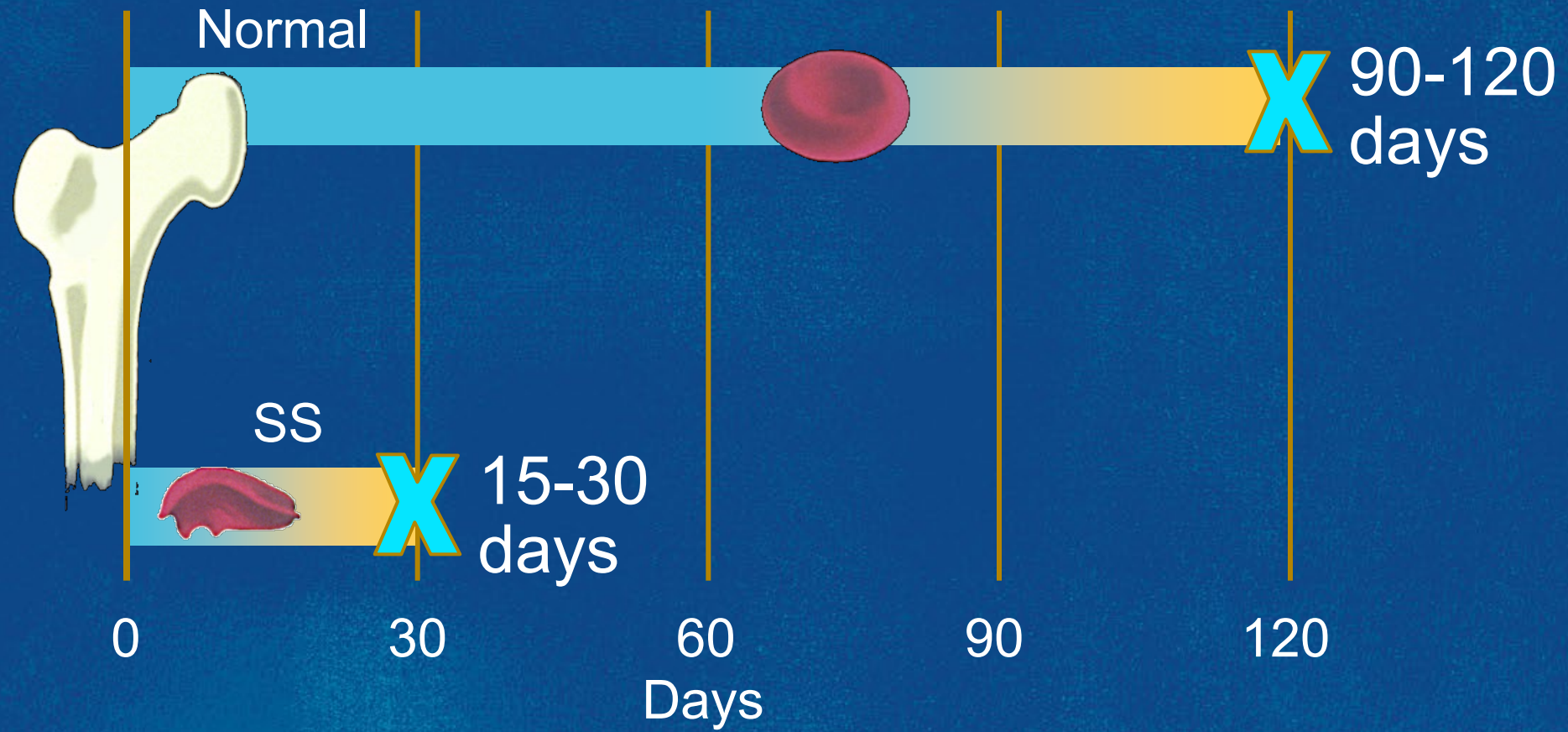
Serving on trial steering committee for Agios

Sickle cell first discovered 120 years ago

- In 1904 - medical intern discovered sickle shaped red cells in the peripheral smear of a dental student from the West Indies



The Lifespan of Sickle RBC is Reduced



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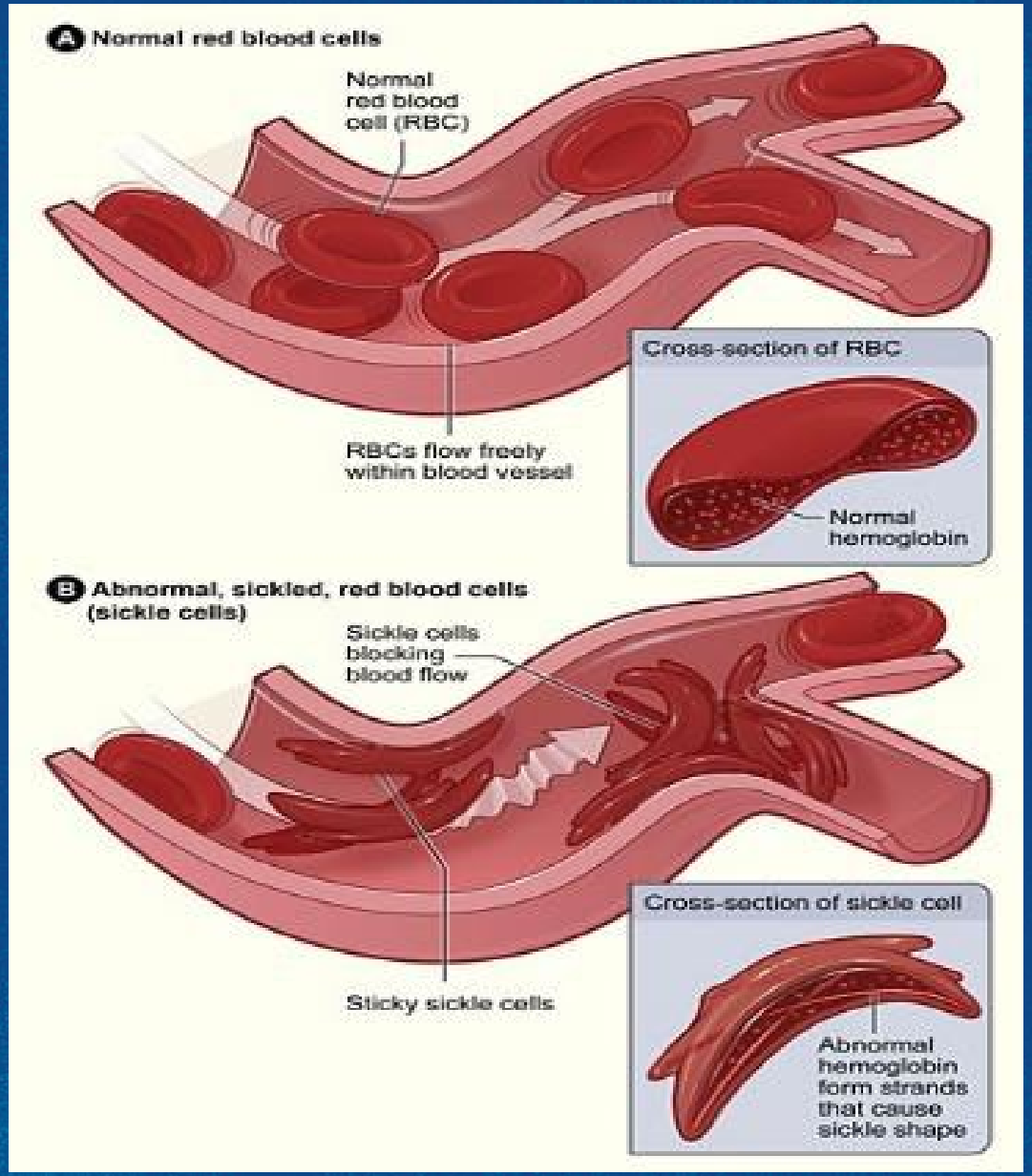
Vaso-occlusion and Pain

- Significant organ damage
- Life expectancy 20 years shorter



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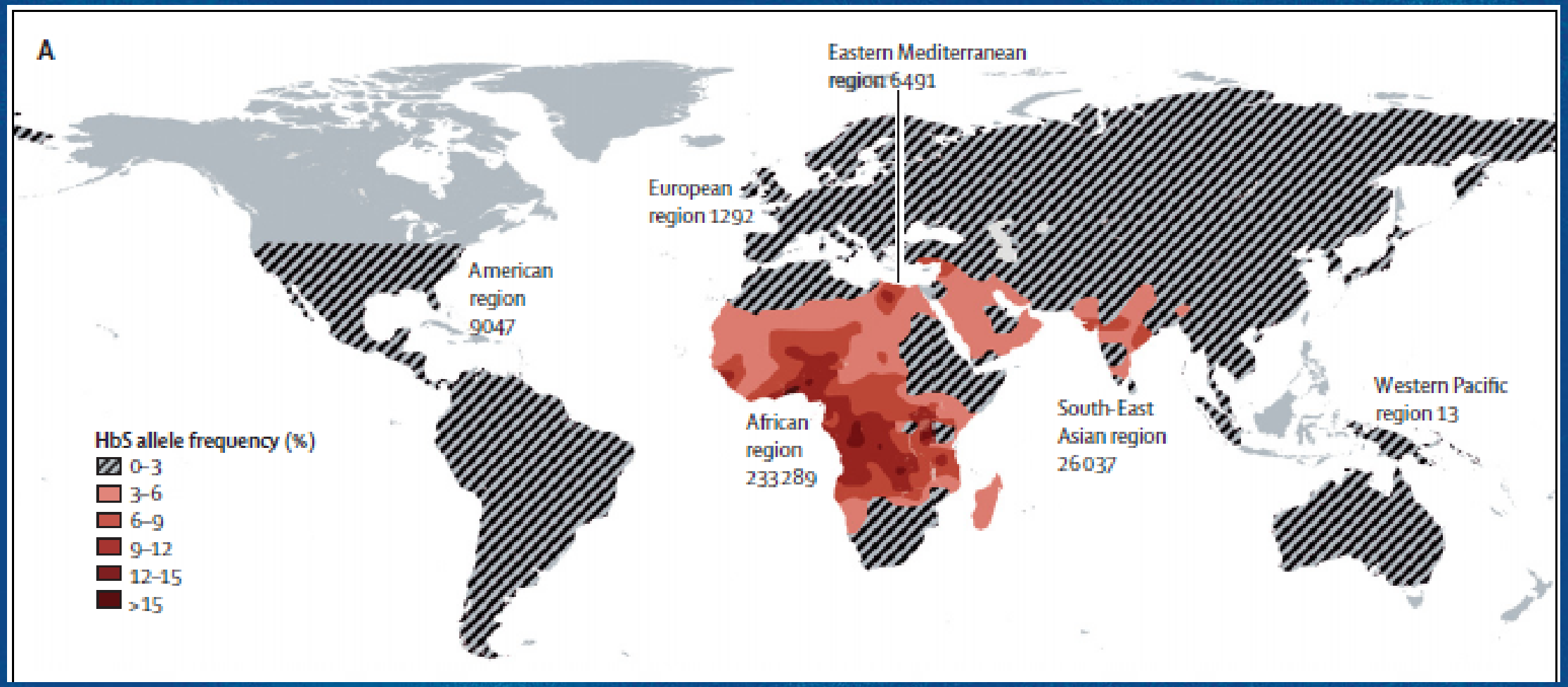
Molecular basis discovered (1949)

- In 1949, Linus Pauling et al. used protein electrophoresis to demonstrate that SCD is caused by a molecular abnormality in the hemoglobin protein
- Established the genetic basis of the disease
- First time a genetic disease was linked to a mutation of a specific protein

World Distribution of SCD gene



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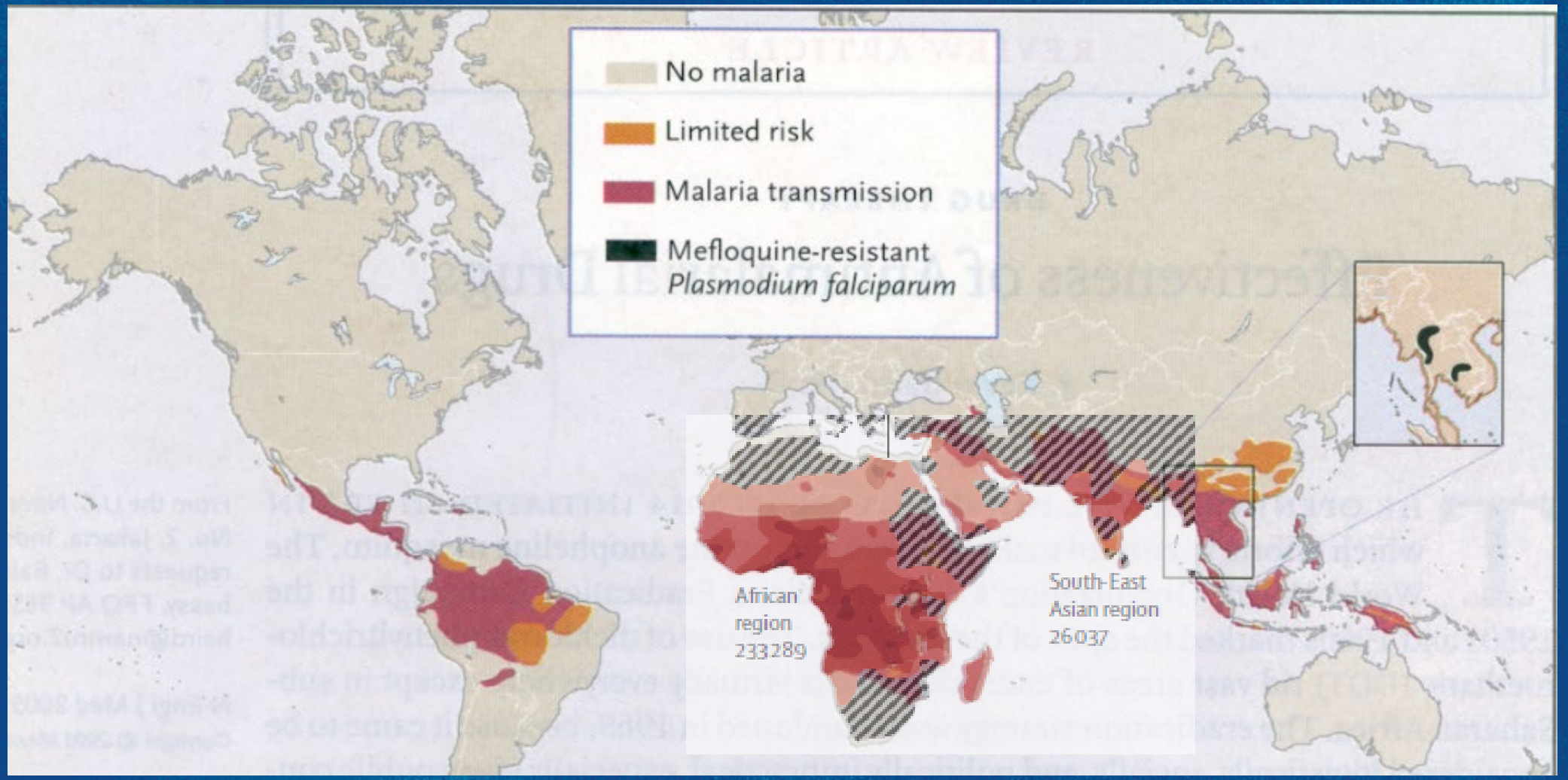
World Distribution of Malaria



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Civil Right Movement

- The Civil Rights Movement highlighted racial disparities in healthcare, prompting increased attention to sickle cell disease

Newborn screening (1970s)

- Method developed for newborn screening for sickle cell disease using blood spots on filter paper, leading to widespread implementation of newborn screening programs across the US

National Sickle Cell Anemia Control Act (1972)

- This act established federal programs for sickle cell education, counseling, research, and voluntary screening, marking a significant step towards widespread testing.

Controversial state laws

- During the 1970s, several states enacted mandatory sickle cell screening laws primarily targeting African Americans, leading to concerns about discrimination against carriers of the sickle cell trait.

Negative social implications

- Due to inadequate education about the difference between sickle cell trait and sickle cell disease, many African Americans faced discrimination in employment, insurance, and other areas based on their carrier status

SCD screening in practice

- Screening is voluntary
- Newborn screening → parental screening
- Proper counseling and education to mitigate potential negative social impacts
- Still limited knowledge in the community


Supacell
a superhero series focusing on young black people in London with latent powers connected to sickle cell disease.

Disparities in clinical research

Minority representation in clinical trials

- White Americans represent 60.7% of the US population
- 90% of the population in clinical trials
- less than 15% of studies report on SES

Alegria M, et al. Reporting of Participant Race, Sex, and Socioeconomic Status in Randomized Clinical Trials in General Medical Journals, 2015 vs 2019. *JAMA Netw Open.*2021
US Census Bureau Quickfacts: United States 2018 [internet]. Washington, DC: Department of Commerce [cited August 20, 2019].

Summary: Improve genomic testing in diverse communities

- Consider the risks
- Education and awareness programs
- Inclusive clinical trials → inclusive clinical guidelines