



BLOOM SYNDROME REGISTRY

Translating research developments to persons with Bloom syndrome

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Bloom Syndrome Registry

Natural history study

Medical concerns

Surveillance

Outcomes


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ORIGINAL ARTICLE

Health supervision for people with Bloom syndrome

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FACE2GENE

Developed by FDNA

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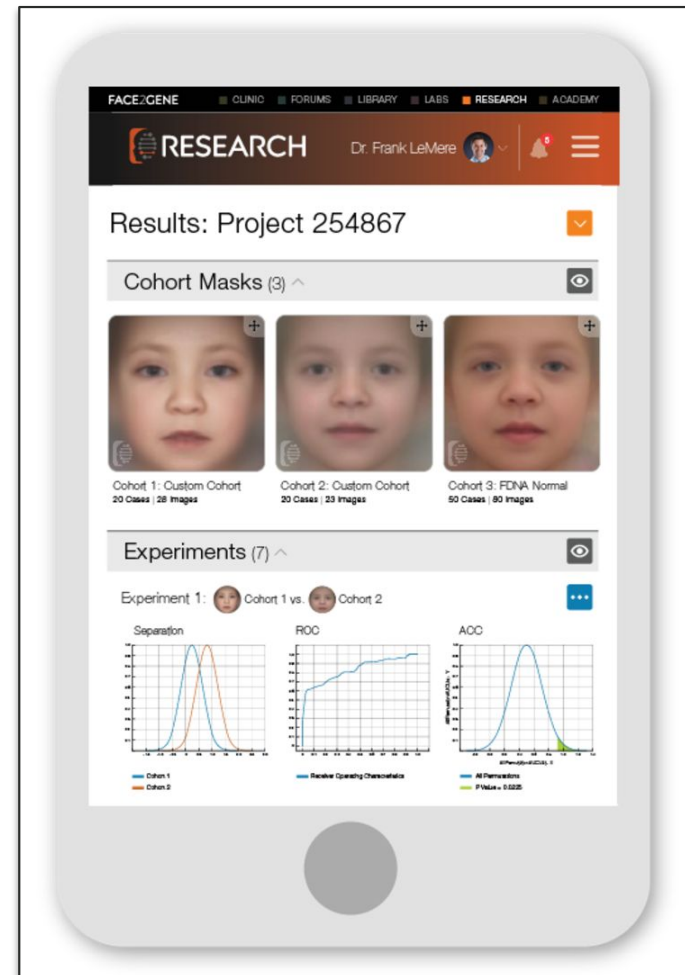


Digital phenotyping

Computer recognition software analyzes facial features

Composite image created

Image analysis by age, gender, ethnicity, genotype



Analysis of tumors

WGS of Bloom tumors has not been reported

The mutation landscape of some tumors implicates
DNA repair pathways

Use the signature to identify therapeutic targets

Currently have 8 tumor/normal samples

Parent / child trios for WGS

Dr. Vivian Chang, UCLA

What are the gene changes in people with Bloom syndrome?

Do these changes affect cancer-related genes?

Surveillance effectiveness

Semi-annual survey of registrants

Is surveillance able to detect cancer early?

Is surveillance able to improve outcome?

Important questions

Effectiveness of cancer treatment

Drug testing

Prevention

Medical and Scientific Advisory Board

The MSAB is committed to furthering the Bloom Syndrome Association's mission to establish a global support network to promote advocacy, stimulate research, and provide education for all members of the Bloom syndrome community.

Medical and Scientific Advisory Board

Chris Cunniff

Nathan Ellis

Joanna Groden

Vivian Chang

Edith Schussler

Maria Vogiatzi

Dermatology

Cancer surveillance

Medical and Scientific Advisory Board

- Provide input to BSA on topics of interest
- Available to answer member questions
- Consider researchers' requests for participation
- Plan for future research