

Patient Samples Updates

NDF Webinar

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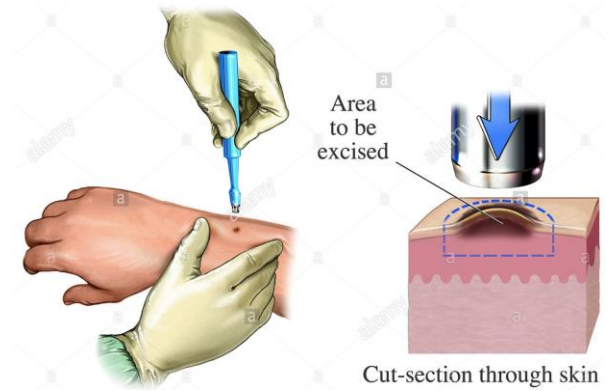


Biospecimens to collect

Blood



Skin biopsy

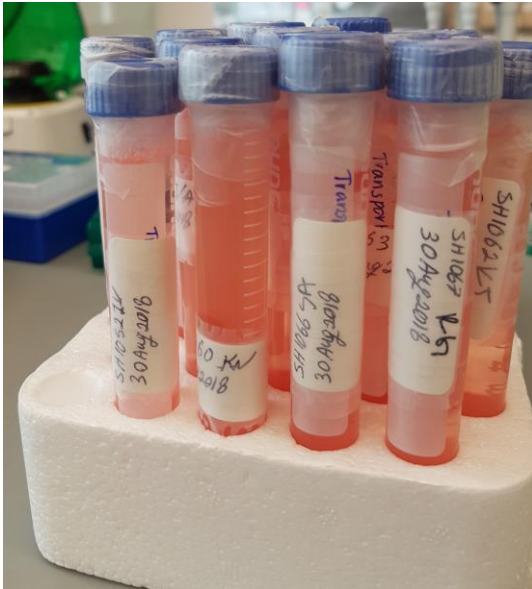


Patient Biobank

1. Skin Biopsy
 - Extract RNA
 - Cell lines
2. Blood sample
 - Extract genomic DNA

Affected patients and their first degree family members (siblings, parents or children)

Received the samples at Yale

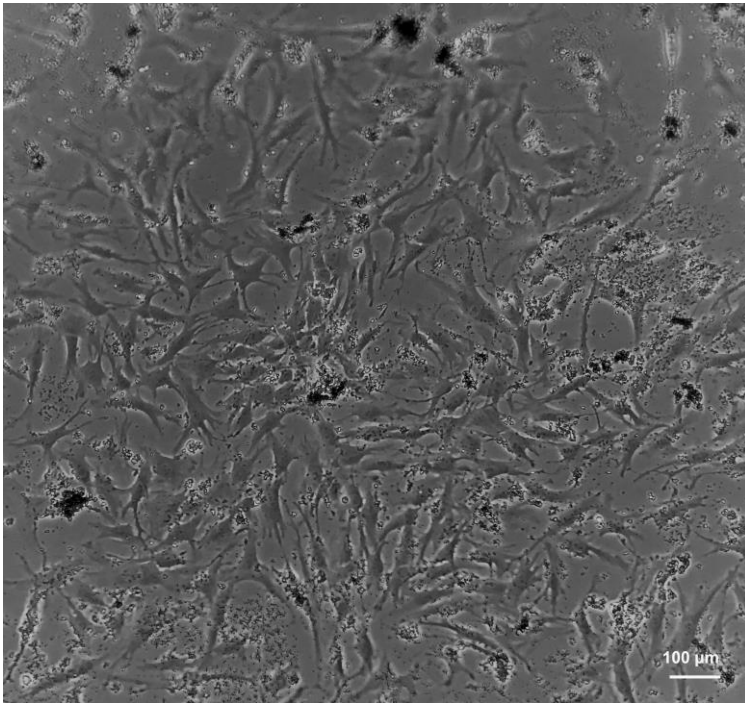


13 skin biopsies

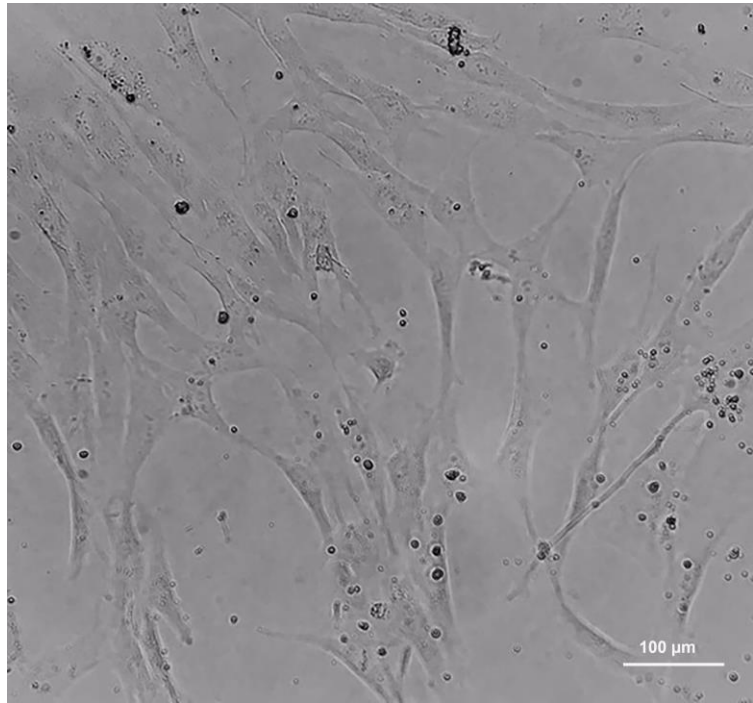


20 blood samples

Patient fibroblast cells created – from half of skin biopsies



SH1062 10x



SH1053 20x

Sitting in the freezer – waiting to be processed



RNA extraction from skin biopsies

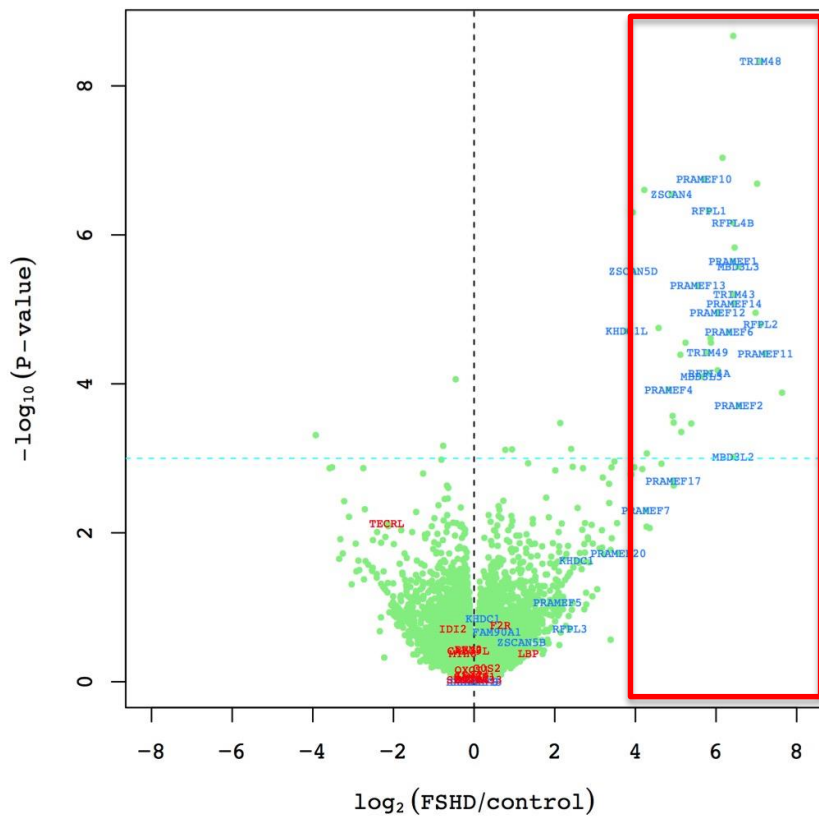


DNA extraction from blood

Next steps

- Obtain extra details on patient donor
- RNA extraction from skin biopsy
 - RNA sequencing and identification of differences between patient and healthy samples
- Extraction of DNA from blood
 - Potential samples for the whole genome sequencing project
- Cell lines from skin biopsy
 - Sequencing validation of GNE mutation
 - Creation of muscle-like cells and other cell types
 - Testing of gene editing approaches

Genetic biomarkers in FSHD



Common FSHD biomarkers:

ZSCAN4 *

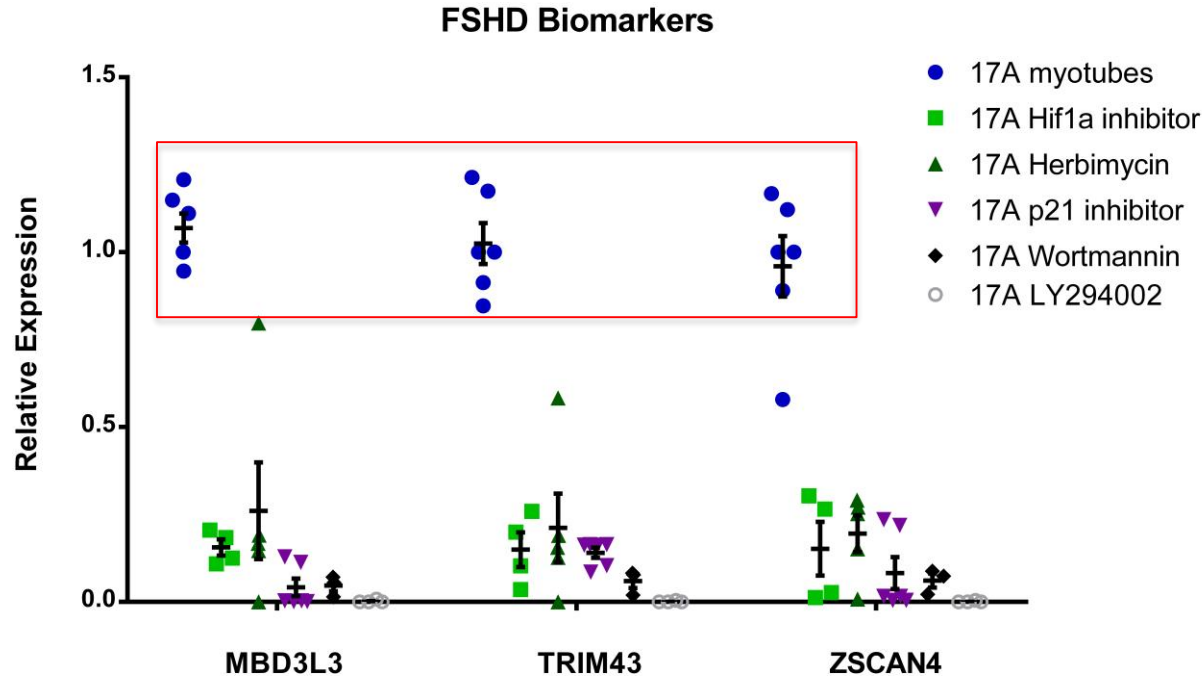
MBD3L3 *

TRIM43 *

PRAMEF1

SLC34A2

Genetic biomarkers can be used to test drugs



Thank you!

- **All patients and family members that volunteered**
- UCLA
 - **Dr Perry Shieh**
 - Kianoush Kamali
- NDF
 - Lale Welsh
 - Nancy Lurie
- Yale
 - **Dr Keryn Woodman**
 - **Dr Heather Best**
 - **Dr. Angela Lek**

Questions

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