

How patient advocacy informs innovation in rare diseases diagnosis

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The stories you are about to hear are from individuals and patient advocates that Illumina works with. The Illumina Global Patient Advocacy team works with patients, families, carers, and the groups that represent them in order to build evidence and advocate for the positive impact of genomics utilization. Patient advocates may be compensated for their time when sharing their stories. Their stories are a testimonial of the potential impacts and benefits genomics can have on disease. One person's experience is not predictive of results in all disease cases, which may differ based on a variety of factors. Results in other cases may vary.

The value of diagnosis

Historically,

- As a mark – stigma
- Research and serendipitous discovery
- Orphan Drug Act 1983
- Language of Endings and Beginnings

Dia - *gignōskein*



Where it all began...



Cambridge Rare Disease Network presents

RARE

summit | 2019

PATIENTS AS PARTNERS

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Understanding the challenge, together

Feb 2020

- Talking
- Listening
- Researching the literature



Meet our unravel Champions

We are really lucky to have a core group of individuals living with r(20) syndrome and parent/carers who are helping us to shape UNRAVEL. Get to know them a little better...



Steve
Baumley
(USA)



Junko
Okuda
(USA/
Japan)



Andre
Reis e
Silva
(Portugal)



Tim
Buckinx
(Belgium/
Netherlands)



Maryse
Barette
(France)



Arantxa
Miralles
(Spain)



Stefan
Grutschus
(Germany)



Yvonne
Goodson
(UK/
Australia)



Dale
Ward
(UK)

Ably supported by...

Laura is our native Italian speaking volunteer who helps us connect families in Italy.

Kalif volunteers supporting our young adults with an r(20) diagnosis, through the Step Into The Ring group (SITR).



Laura
Giordano
(Italy)



Kalif
Garticki
(SITR)



Building a team...

- Feb 2021
- Ring20 champions
- Families, helping families
- Getting everyone involved



UNRAVEL

Making genomic understanding of Ring Chromosome 20 Syndrome possible

Welcome to our UNRAVEL project page!

Here you'll find all the information about our Ring20 collaborative project with Illumina Inc.

Why UNRAVEL?

Ring Chromosome 20 Syndrome is a rare and under researched condition, raising many questions from families and clinicians/scientists alike. Ring20 are embarking on a journey to try to UNRAVEL some of the mysteries of r(20) to see if we can make diagnosis easier and find clues to help with better treatments in the future.

UNRAVEL comprises two distinct phases:

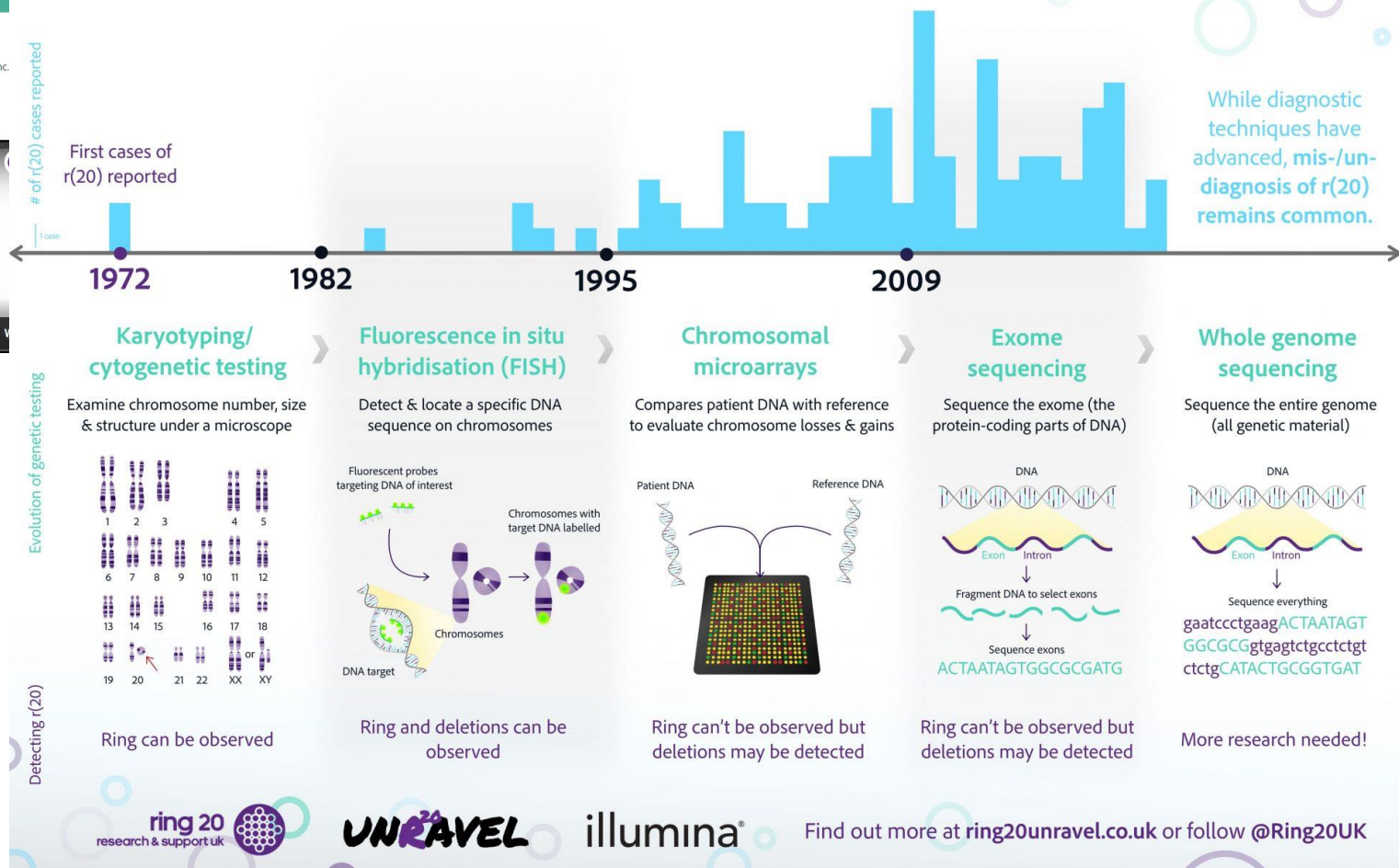
- PHASE I comprises a patient family education program, alongside preparations for research
- PHASE II comprises patient recruitment for a research and development study

mosaic

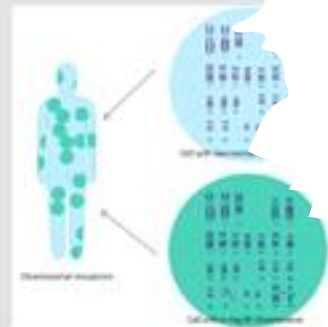
Orange = r(20) cells
Blue = normal cells



Evolution of genetic testing & its impact on r(20) diagnosis



r(20) means... you have a Ring chromosome 20



READ MORE ABOUT PARTS OF A CHROMOSOME



Hosting our 2nd ever Families Conference

September 2022

- Explaining genomic sequencing and how it might help
 - The ring is merely a 'symptom' – we need to find the root cause
- Lab tours
- Strawberry DNA extraction





It takes a village...

How to get samples from patients to industry?

(solved by) February 2023

- Industry proposing R&D
- Patient Community willing to participate

A collaborative solution required...





Our journey so far...



RAREsummit19/
Lily's interview
Sep 2019



Ring20
Champions
formed
Feb 2021

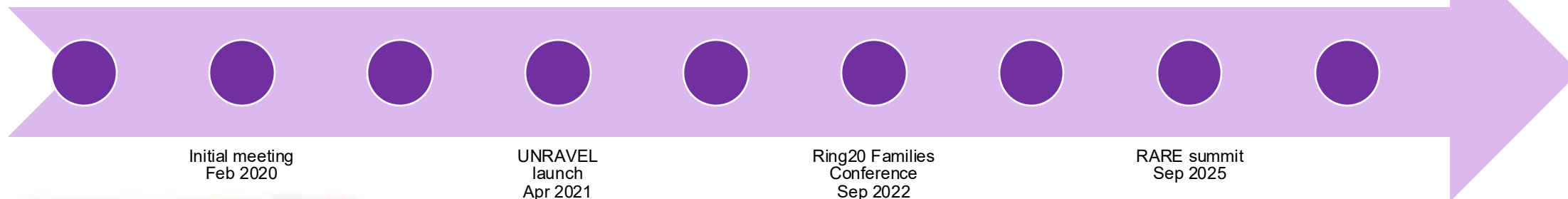


Ring20
animation
Feb 2022



Genomic Pilot
Study
commenced
Feb 2023

The future...?



Initial meeting
Feb 2020

UNRAVEL
launch
Apr 2021

Ring20 Families
Conference
Sep 2022

RARE summit
Sep 2025



The Undiagnosed Hackathon

Pioneered in 2023 by the Wilhelm Foundation, and PhenoTips

Brings together the broadest possible array of tools alongside clinicians, bioinformaticians, molecular biologists, scientists, developers, AI specialists, and more to collaborate in solving some of the most difficult patient diagnoses globally.



Thank you

Shirlene Badger

Illumina