



L O U D R A R E



NADINE
LIVING WITH FIBRODYSPLASIA
OSSIFICANS PROGRESSIVA
BERLIN, GERMANY

Insights into my rare disease journey as a patient, patient advocate and researcher



Nadine Z. Großmann, M.Sc.

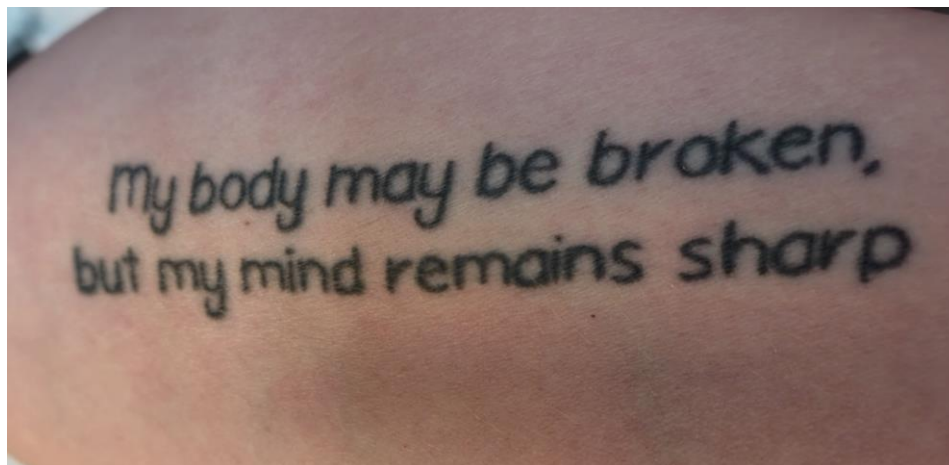
Vice chair FOP Germany

Vice chair IFOPA's board of directors

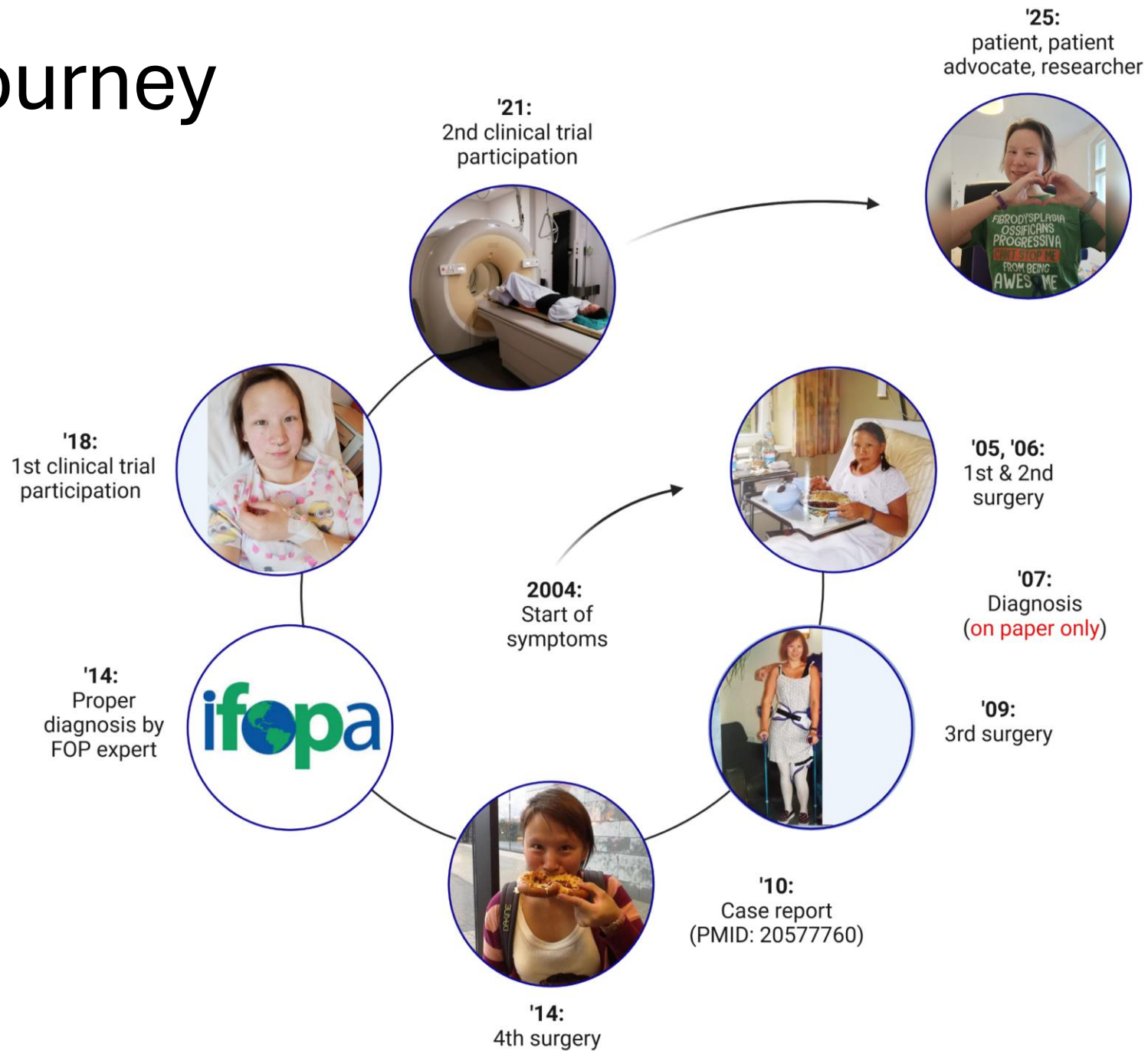
Ph.D. candidate



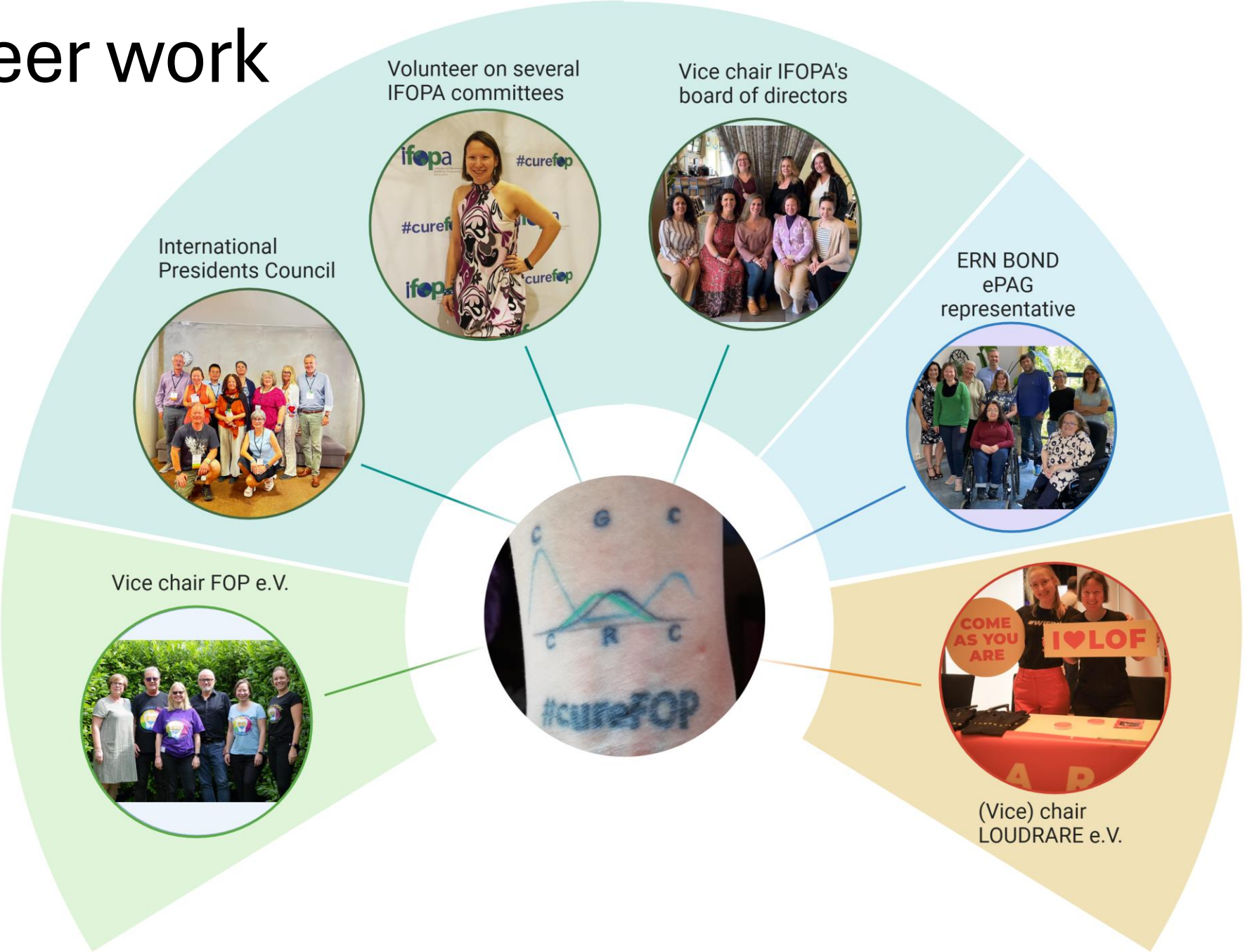
Explaining FOP Through Tattoos



My FOP journey



Volunteer work



FOP Germany

- Founded in 1998
- >200 members
- 48 German FOP patients
- Our programs:
 - Support FOPers and their families
 - Public outreach/raise awareness
 - Production and dissemination of educational material
 - Health policy
 - Collaboration with involved stakeholders
 - Fundraising
 - Support research worldwide



FOP e.V. Jahrestagung 2024

FOP documentary

- Videographer visited Sarah and her parents every 2-3 months over 2 years
- What is it like to live with an ultra-rare disease?
- What is the current state of drug development?
- If Sarah had one free wish, what would it be?

Ein Dokumentarfilm über eine der seltensten Erkrankungen der Welt

OFFICIAL SELECTION
FILMTAGE
OBERSCHWABEN
2024

Prädikat
wertvoll
FBW

bis auf die
Knochen

Leben mit **FOP**

May 13th, 8.30 pm
Casino Aschaffenburg

EIN FILM VON MICHAEL SCHEYER
© 2024 ATELIER FÜR SEHENSWERTE MEDIEN
lebenmitfop.de

FÖRDERUNG DES KINOVERTREIBS
IPSEN

Loudrare e.V.

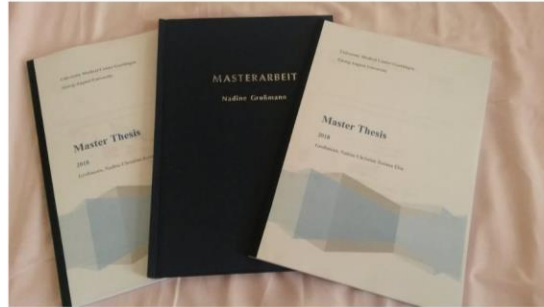
- Founded in 2022 by 5 young people living with rare diseases
- Developed **award-winning Germany-wide awareness campaign** for people living with rare diseases - Mensch #wiedu ("People #likeyou"), biggest and loudest of its kind in Germany
- Created a **digital community** on social media, enabling those affected to actively engage, such as raising awareness about rare diseases
- We host the **Loudrare Online Festival**, where individuals can connect with each other and experts
- The voices of 4 million affected individuals in Germany must be heard. This is key to greater acceptance, earlier diagnoses, dedicated doctors, better education, access to information and therapies, and reduced stigma and discrimination

→ Make some noise so that rare someday becomes normal



My FOP researcher career

Master Thesis:
Characterization of a potential
biomarker to predict flare ups
in patients with FOP



Ph.D. Thesis:
Age-dependency in
health and the ultra-
rare disease FOP

Hypothesis:

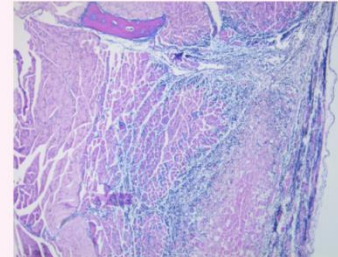
Age effects on the onset and progression of HO in FOP are due to a **dampened immune response** and **altered skeletal muscle regeneration capacity** in adults resulting in milder and less robust formation of HO with age

Goal

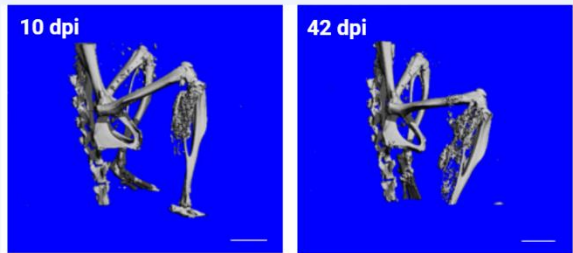
Determine age-dependent progression of HO and tissue repair in FOP over time



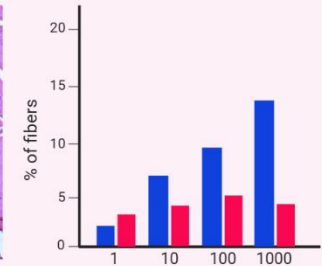
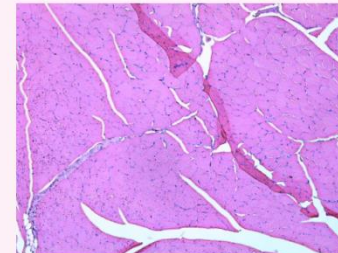
microCT



Histology



in vivo microCT



Quantification of cross sectional area

Thank you!

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📷 @one.raregirl, @fop_germany,
@loudrare

🌐 Nadine Z. Grossmann



Challenges I face(d) living with an ultra-rare disease

- Misdiagnosis
- 4 risky and contra-indicated surgeries
- 3 years until initial diagnosis
- 10 years until finding an FOP expert and FOP patient orgs
- Tons of additional paperwork
- Fighting rejections
- Constantly need to advocate for myself (esp. when seeing new doctors)
 - Medical gaslighting
- Social exclusion/isolation
- Discrimination and being stigmatized
- Ableism
- Unsolicited „advice“

Genetic testing in FOP

- 3.5 year old boy gets injured while playing and develops a **tissue swelling** (flare-up)
- As the swelling travels doctors perform an MRI and take a **biopsy** and don't find anything
- Swelling continues to grow and travel
- Doctors believe it's a **tumor**
- Parents do some internet research and find **FOP**; for them, all the puzzle pieces fall into place
- Same day: 2nd evaluation of biopsy returns: **benign tumor**
- Parents inform doctors about FOP and emphasize they'd like to have **genetic testing** done
- Doctors don't believe in their „theory“
- Parents end up sending all documents **on their own** to German FOP expert



9.12.24

10.12.24