MD-PhD Candidate Sophie Huisman

Leiden University Medical Center

‘Outcomes of the FD/MAS module’

EuRREB
European Registries for Rare
Endocrine and Bone conditions

The FD/MAS module – Internship to PhD

Drs. Sophie A. Huisman

Leiden University Medical Center (LUMC)

The Netherlands

EuRREB
European Registries for Rare
Endocrine and Bone conditions

Funded by
the European Union

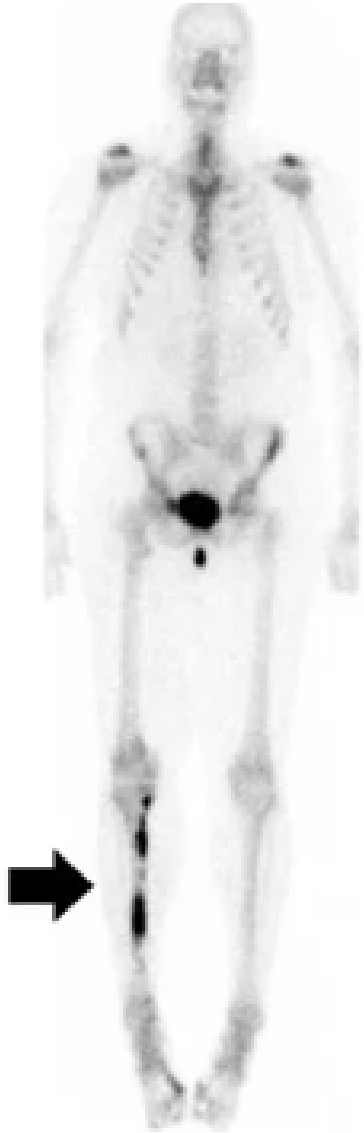
My internship – March 2024 to August 2024

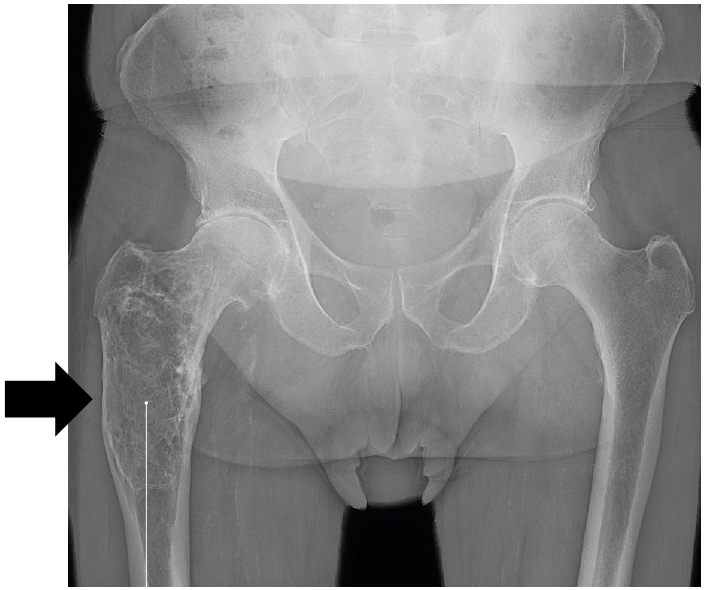
- Module training and data entering at the LUMC
- Data entering at Oxford and Bochum

- **Practical tips for supervisors**
  - Have patient lists available and make sure your student can access the medical files
  - Make sure patients have signed the informed consent so your student can start straight away
  - Let student arrange housing in time (Facebook groups)
  - Discuss possible travel grants with your students
  - Medical language is often the same in many countries, students will pick it up easily

Fibrous Dysplasia/McCune Albright Syndrome (FD/MAS)

- GNAS gene
- Skeletal lesions
- Hyperpigmented skin lesions
- Myxomas
- Endocrine disorders
  - Precocious puberty
  - Hyperthyroidism
  - GH overproduction
  - Hyperprolactinemia





FD/MAS in e-REC (n=3097)

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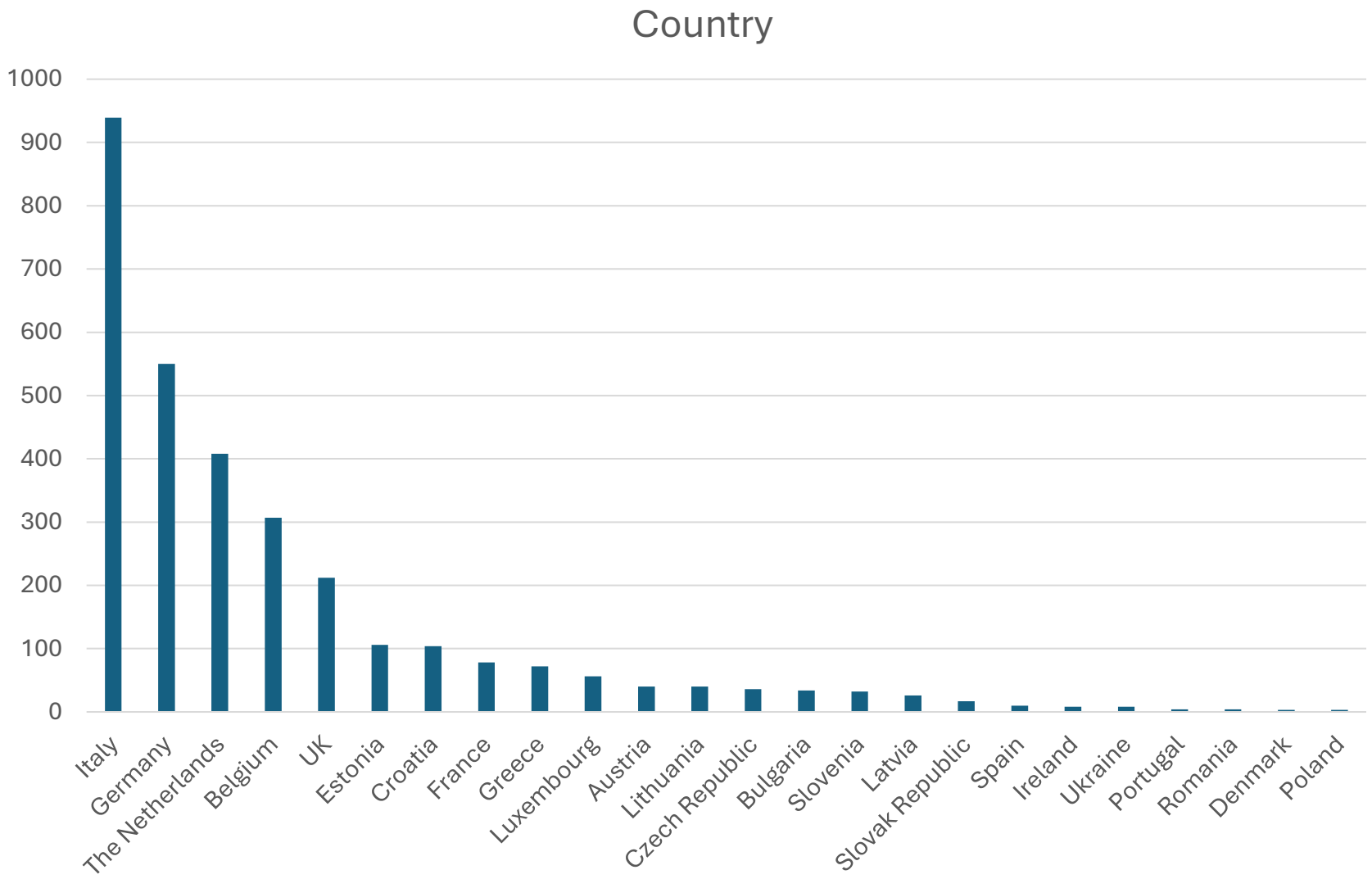
e-REC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Greece
Luxembourg
Austria
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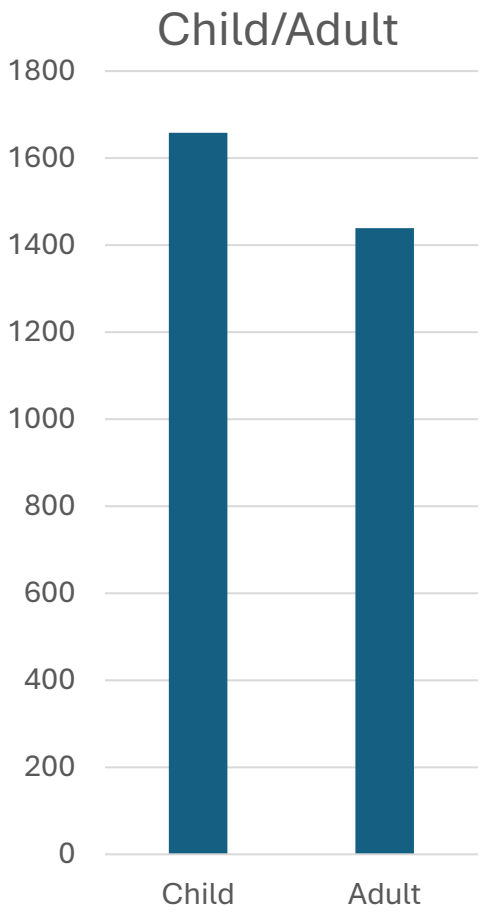
Latvia
Slovak Republic
Spain
Ireland
Ukraine
Portugal
Romania
Denmark
Poland


Child/Adult
1800
1600
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Child
Adult

FD/MAS in Core Registry (n=696)

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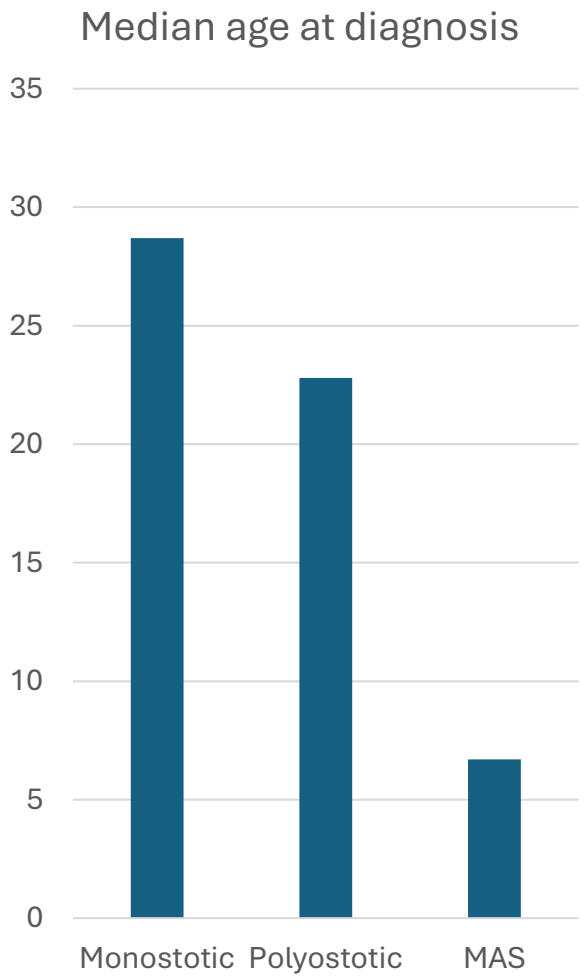
Monostotic FD diagnosis at older age

- Adults (n=639)
  - 62.9% Female
  - 49% Monostotic, 25.7% Polyostotic, 12.7% MAS
- Children (n=57)
  - 54.4% Female
  - 29.8% Monostotic, 26.3% Polyostotic, 36.8% MAS



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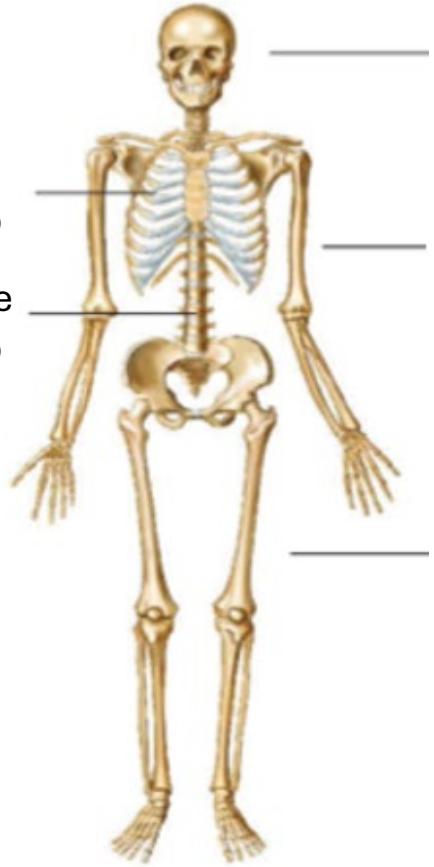
- Median diagnostic delay 1.3 years (IQR 0.4 to 4.6)

	% of MAS	Mean age of onset (years)
Precocious puberty	56.4%	4.4 (SD 2.6)
Hyperthyroidism	11.5%	18.6 (SD 12.8)
GH overproduction	9.0%	19.3 (SD 17.2)
Hyperprolactinemia	9.0%	21.9 (SD 13.5)

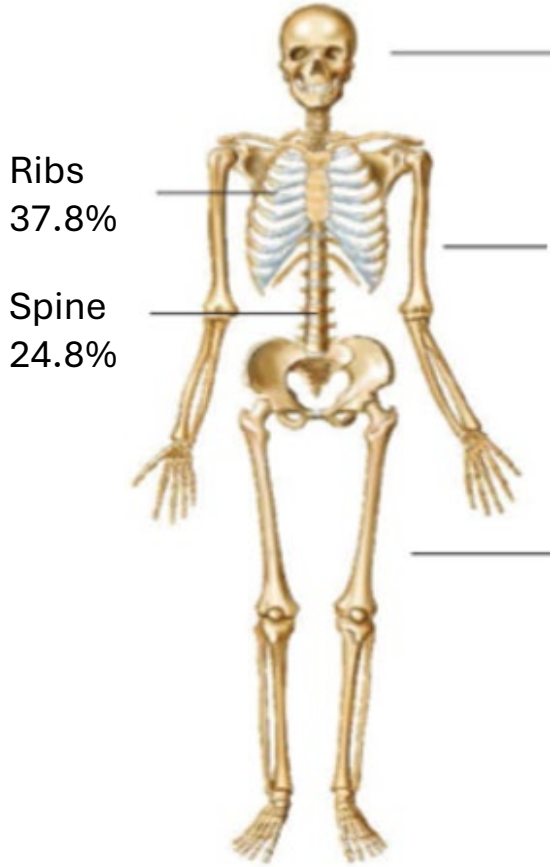
First presentation of FD/MAS in expertise center

FD location

Monostotic



Polyostotic



Clinical characteristics at first presentation

Adults	MFD (n=175)	PFD (n=108)	MAS (n=34)
Pain	57.9% Data available 159/175	73.3% Data available 86/108	84.6% Data available 26/34
Previous fractures	9.5%	46.6%	70.6%
Bone Turnover Markers (Ctx, P1NP, AF)	0 high: 52.7% 1 high: 30.4% 2 high: 12.5% 3 high: 4.5% Data available 112/175	0 high: 19.7% 1 high: 31.1% 2 high: 27.9% 3 high: 21.3% Data available 61/108	0 high: 7.7% 1 high: 30.8% 2 high: 15.4% 3 high: 46.2% Data available 13/34

Children	MFD (n=36)	PFD (n=17)	MAS (n=11)
Pain	36.7% Data available 30/36	60.0% Data available 10/17	55.6% Data available 9/11
Previous fractures	22.2%	53.3%	55.5%
Bone Turnover Markers (Ctx, P1NP, AF)	0 high: 13.0% 1 high: 26.1% 2 high: 47.8% 3 high: 13.0% Data available 23/36	0 high: 16.7% 1 high: 16.7% 2 high: 33.3% 3 high: 33.3% Data available 6/17	0 high: 0% 1 high: 0% 2 high: 0% 3 high: 100% Data available 2/11

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Pain

- Monostotic FD
  - Spine (87.5%) and upper extremity (81.8%) most often painful
  - Craniofacial (40.2%) least often painful

- Females
- Polyostotic FD
- MAS
- Previous fractures

- Most important factors:
  - Females: OR 2.57 ($p<0.001$)
  - Polyostotic FD: OR 2.43 ($p=0.001$)

Conclusions

- Collecting data in an international registry is possible, especially with the help of motivated medical students
- For the FD/MAS module there are still many patients (especially children) to be entered in the full module, I am happy to help! (also in training your students)
- MFD is more often diagnosed later in life and most diagnostic delay is before first presentation in an expertise center
- Pain presentation is different in children compared to adults
- Women experience more pain than men, especially when diagnosed with PFD
- Craniofacial least often painful in adults, probably due to the increased incidence of CT scans after the age of 50

Questions?

Thank you

Ways to contact us:

eurreb.eu

registries@lumc.nl

drop-in sessions via Zoom

European Registries for Rare Endocrine and Bone Conditions

EuRREB
European Registries for Rare
Endocrine and Bone conditions

Endo-ERN
European Reference Network on Rare Endocrine Conditions

ERN BOND
EUROPEAN REFERENCE NETWORK ON RARE BONE DISEASES