WHAT THE PATIENT REGISTRY IS FOR ...

- To improve our understanding of the disease of FD / MAS in the patient population.
- To improve our understanding of diagnosis and treatment of FD / MAS.
- To improve our understanding of the patient experience of illness -- how you experience symptoms and your treatment both personally and socially.
THE FDFPR ...

- Consists of a set of 12 interrelated, but independent surveys.
- Completing it takes some time, especially if you have many symptoms, have received extensive surgical and/or medical treatment.
- It can be completed in stages.
- We ask that everyone complete the long process once.
- Follow-up surveys in later years will be shorter.
2 SURVEYS DOCUMENT INFORMATION ABOUT THE PATIENT AND DIAGNOSIS

- basic information about the participant
- diagnosis survey
4 SURVEYS DOCUMENT SYMPTOMS EXPERIENCED BY PATIENTS

- Fractures survey
- Birth, puberty, and reproductive history survey
- Endocrine disorders and other symptoms survey
- Pain level survey
4 SURVEYS DOCUMENT TREATMENTS RECEIVED AND PATIENT SATISFACTION

- skeletal surgeries survey
- pain treatments and other medications survey
- pain treatment follow-up questions survey
- other medications follow-up questions survey
Day-to-day needs:
- How FD/MAS affects your ability to complete activities of daily living
- How FD/MAS affects your ability to fulfill your social roles?

Mental health and well being:
- How FD/MAS affects the patient’s emotional state: feelings about yourself and life (happiness, depression, anxiety, anger, etc.)
- How FD/MAS affects social relations: Whether you face exclusion/inclusion, support/discrimination, etc.
WHAT IS THE USE OF THE FDFPR?

- It enables exploration of trends in the patient population.
- It enables exploration of relationships between specific symptoms and QOL.
- It enables clinical effectiveness research to be conducted so clinicians will have a scientific basis for making specific treatment recommendations.
WHAT IS THE REPRESENTATION OF SYMPTOMS IN THE PATIENT POPULATION?
- What bones are affected by FD?
- Which bones are fracturing?
- How much pain are patients experiencing?
- What kinds of pain are they experiencing?

WHAT ARE THE TRENDS IN DIAGNOSIS:
- When is FD being identified?
- Who is making diagnoses?
- Through what means is diagnosis made?
POPULATION TRENDS QUESTIONS

- **Trends in treatment:**
  - What types of treatments are in use for specific symptoms?
  - Do demographic qualities of patients determine their access to care for specific symptoms?

- **Trends in experience of quality of life**
RELATIONSHIPS BETWEEN SYMPTOMS AND QOL

- Does the number of bones affected by FD correlate with an individual’s quality of life? For adults? For kids?

- Does the kind of pain experienced and/or its intensity and/or its location correlate with quality of life? For adults? For kids?

- Does FD in the bones of the face / skull affect QOL differently than bones in other parts of the skeleton? For adults? For kids?
Does a particular treatment improve the quality of life of the patient?

Which of two treatments result in better treatment outcomes?
- Rods or plates?
- X medication of Y medication?
- Surgery or watchful waiting?
What’s the status of the FDFPR? Currently...

- Patients Registered: 538
- Some surveys completed: 238
- All surveys completed: 75

Please sign up, sign in, and work through the surveys!
## Who is currently in the FDFPR?

<table>
<thead>
<tr>
<th>Age (N=171)</th>
<th>%</th>
<th>Frequency</th>
</tr>
</thead>
<tbody>
<tr>
<td>0 - 9</td>
<td>6%</td>
<td>10</td>
</tr>
<tr>
<td>10 - 19</td>
<td>17%</td>
<td>29</td>
</tr>
<tr>
<td>20 - 29</td>
<td>21%</td>
<td>36</td>
</tr>
<tr>
<td>30 - 39</td>
<td>21%</td>
<td>36</td>
</tr>
<tr>
<td>40 - 49</td>
<td>19%</td>
<td>32</td>
</tr>
<tr>
<td>50 - 59</td>
<td>9%</td>
<td>16</td>
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<tr>
<td>60 - 69</td>
<td>7%</td>
<td>12</td>
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</table>

<table>
<thead>
<tr>
<th>Race (N = 186)</th>
<th>%</th>
<th>Frequency</th>
</tr>
</thead>
<tbody>
<tr>
<td>Asian</td>
<td>4%</td>
<td>8</td>
</tr>
<tr>
<td>Black</td>
<td>1%</td>
<td>2</td>
</tr>
<tr>
<td>Native / Hawaiian</td>
<td>3%</td>
<td>6</td>
</tr>
<tr>
<td>White</td>
<td>89%</td>
<td>165</td>
</tr>
<tr>
<td>Other</td>
<td>3%</td>
<td>5</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Gender (N=180)</th>
<th>%</th>
<th>Frequency</th>
</tr>
</thead>
<tbody>
<tr>
<td>women</td>
<td>79%</td>
<td>143</td>
</tr>
<tr>
<td>men</td>
<td>21%</td>
<td>37</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>County (N = 184)</th>
<th>%</th>
<th>Frequency</th>
</tr>
</thead>
<tbody>
<tr>
<td>USA</td>
<td>85%</td>
<td>156</td>
</tr>
<tr>
<td>Canada</td>
<td>4%</td>
<td>8</td>
</tr>
<tr>
<td>Other</td>
<td>11%</td>
<td>20</td>
</tr>
</tbody>
</table>
### Diagnosis (N=168)

<table>
<thead>
<tr>
<th>Diagnosis</th>
<th>%</th>
<th>Frequency</th>
</tr>
</thead>
<tbody>
<tr>
<td>Monostotic FD</td>
<td>42%</td>
<td>71</td>
</tr>
<tr>
<td>Polyostotic FD</td>
<td>37%</td>
<td>62</td>
</tr>
<tr>
<td>Monostotic &amp; MAS</td>
<td>3%</td>
<td>5</td>
</tr>
<tr>
<td>Polyostotic &amp; MAS</td>
<td>18%</td>
<td>30</td>
</tr>
</tbody>
</table>

### Commonly Affected Bones (N = 169)

<table>
<thead>
<tr>
<th>Bone</th>
<th>%</th>
<th>Frequency</th>
</tr>
</thead>
<tbody>
<tr>
<td>Skull</td>
<td>45%</td>
<td>73</td>
</tr>
<tr>
<td>Mandible</td>
<td>17%</td>
<td>28</td>
</tr>
<tr>
<td>Left Femur</td>
<td>33%</td>
<td>56</td>
</tr>
<tr>
<td>Right Femur</td>
<td>25%</td>
<td>42</td>
</tr>
<tr>
<td>Left Tibia</td>
<td>22%</td>
<td>37</td>
</tr>
<tr>
<td>Right Tibia</td>
<td>20%</td>
<td>33</td>
</tr>
<tr>
<td>Pelvis</td>
<td>18%</td>
<td>30</td>
</tr>
</tbody>
</table>
Planning is under way for a Clinical Effectiveness study concerning FD in the face, skull or skull base.

Does surgery produce better QOL outcomes for individuals with Craniofacial FD (CFD) than “doing nothing”?

Do CFD patients who receive surgery have:
- Better physical functioning?
- Less frequent pain? Less intense pain?
- Fewer emotional difficulties?
- Better social functioning?
AN INVITATION TO PARTICIPATE:

- Individuals with FD in face, skull and skull base are strongly encouraged to register in the FDFPR and complete the key surveys ASAP.

- https://www.fdmasregistry.org
CRANIOFACIAL CER STUDY - KEY SURVEYS

- basic information about the participant diagnosis
- skeletal surgeries
- pain level
- mental health and well being
- day to day needs
- Fractures
- birth puberty and reproductive history
- endocrine disorders and other symptoms
- pain treatments and other medications
- pain treatment follow-up questions
- other medications follow-up questions
- clinical documents (an opportunity to upload documents, not so much a survey)
Everyone with rare disease benefits when individual patients put their information together.

We can understand our disease better, we are able to improve treatment, and we will do a better job of making it possible for more people to live well with symptoms.

The FDFPR is only as good a tool as we make it. Numbers matter and quantity of data matters. If we had 538 complete sets of data instead of 70, we’d be in an amazing position to answer questions!

Your input is needed!
Research about the Patient Experience of Illness and Coping Strategies

- Open ended interviews help us better grasp how patients and caretakers make decisions about care and negotiate daily life and social situations.

- Qualitative research can be useful to help develop CER studies.

- I am conducting a qualitative investigation of patient coping and surgical decision making
Individuals 20+ years old with FD in their face, skull, or skull base are encouraged to contact me if they are interested in completing an open ended interview about their coping and surgical decision making (if relevant):

- AKONRADI@LOYOLA.EDU

- Subject line: CFD study