

Response Evaluation In Neurofibromatosis Schwannomatosis
INTERNATIONAL COLLABORATION

- If sharing any data or information from these slides generated by the REiNS International Collaboration, please acknowledge the authors, group chairs, and specific working group.
- If using any information presented with a citation, please reference the primary source.



REiNS Functional Group Update

June 17, 2022

Andrea Gross, MD &



Response Evaluation In Neurofibromatosis Schwannomatosis
INTERNATIONAL COLLABORATION

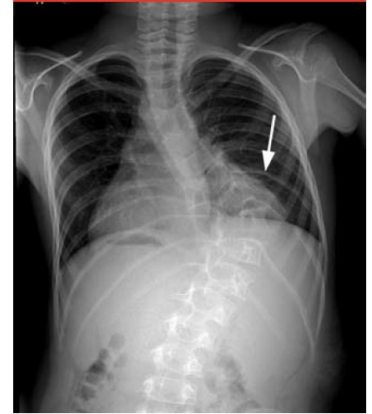
Functional Group Update

- Skeletal Manifestations of NF1
 - MRI vs Standing Radiograph for measurement of scoliosis in NF1
 - NF1 Bone Manifestations Landscape Manuscripts
- Disfigurement Rating Scale
- Future Plans for Functional Committee



Scoliosis in NF1: X-Ray vs MRI

- Standing Radiograph (X-Ray) is the gold standard for measuring scoliosis angle
- Magnetic Resonance Imaging (MRI):
 - Standard imaging modality for NF related tumors
 - Can visualize progression of scoliosis on MRI
 - Uncertain relationship between scoliosis curve measured on radiograph vs MRI



AP radiograph demonstrating rib penciling (arrow) in a patient with neurofibromatosis-1. (Courtesy of Lori A. Karol, MD, Texas Scottish Rite Hospital, Dallas, TX.)

04-2003

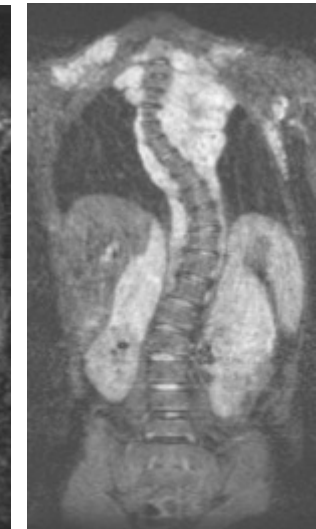
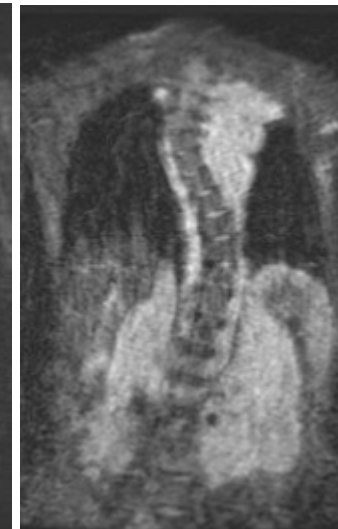
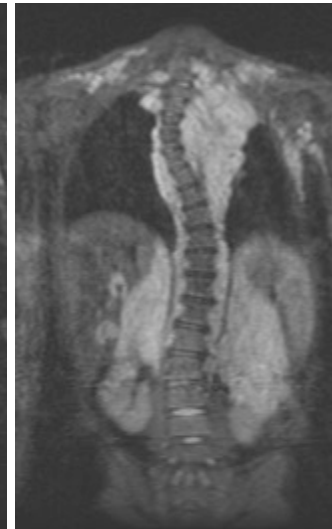
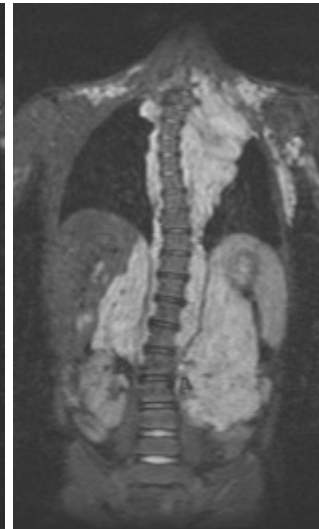
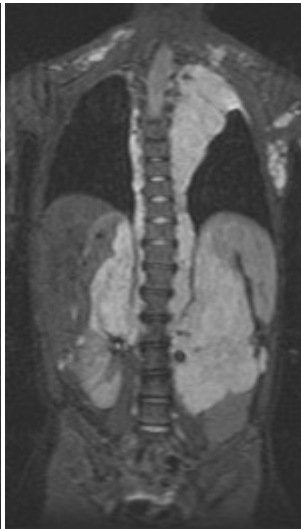
02-2004

04-2005

04-2006

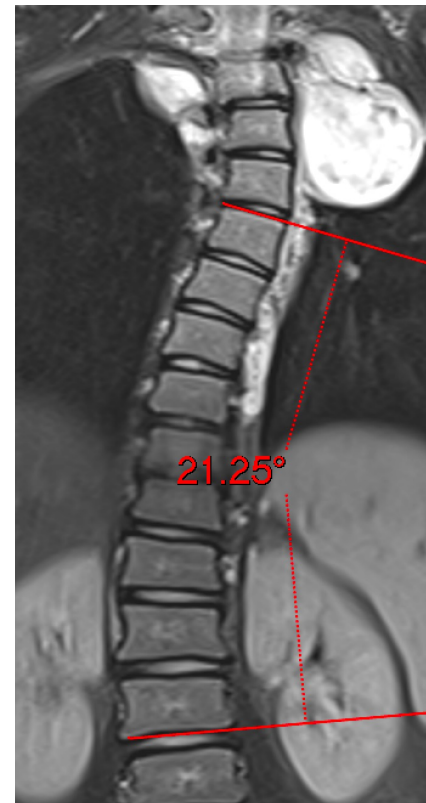
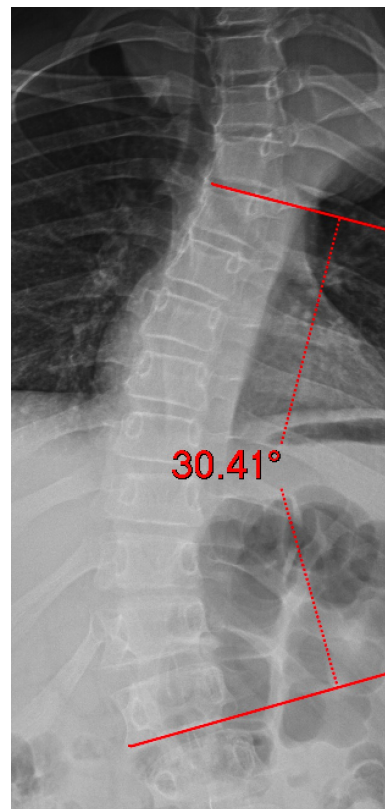
09-2006

02-2007



Hypotheses

The degree of spinal curvature and changes in degree of curvature visualized on MRI can be directly correlated with the degree of spinal curvature seen in standard standing scoliosis radiographs (Cobb angle) obtained at the same time point



Study Design & Eligibility

- Design: Retrospective review comparing Cobb angles of MRI scans and standing radiographs of the spine obtained on patients as part of clinical care or a previous clinical trial.
- Eligibility: Patients (any age) with a clinical or genetic diagnosis of NF1 who have an MRI which includes the entire spine (cervical, thoracic and lumbar) and standing scoliosis radiographs completed within 3 months of each other



Scoliosis Study Implementation

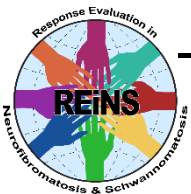
- ☑ Independent Radiologist Readers identified
- ☑ Feedback from REiNS Community (March 2021 Meeting)
- ☑ Protocol submitted by Dr. Laura Klesse to UT Southwestern IRB by Dr. Laura Klesse
- ☑ Protocol approved by IRB
- ☐ Establishing Radiology Research database to be able to collect + deidentify eligible scans and calculate MRI and radiograph Cobb angles



NF1 Skeletal Endpoints Landscape

Manuscripts

- Committee plans to write 3 manuscripts:
 - Overall Bone Health in NF1
 - Focus on Osteopenia/Osteoporosis
 - Scoliosis
 - Long Bone Dysplasia/Pseudoarthrosis
- For each paper, will present:
 - Available measurement techniques/markers for each condition
 - Review of use of these techniques in clinical trials (NF and other)
 - Identification of areas in need of additional research



Disfigurement Rating Scale

- External Rating Scale Developed by NCI Team (Dr. Liny John)
 - Input from REiNS Patient Representative Committee part of development process
- Presented to REiNS Functional Committee in May 2022
- Needs additional prospective data/evaluation
- Plan to collaborate with PRO Group to compare against patient-reported disfigurement related distress rating scale



Functional Committee Future Plans

- Gastrointestinal symptoms in NF1
 - Andres Lessing
- Revisiting/revising recommendations from previous publications
- Other??



Thank you!!

- REiNS Functional Committee Members:
 - Andres Lessing
 - Bev Oberlander
 - Brigitte Widemann
 - Brittany Simpson
 - David Stevenson
 - David Viskochil
 - Gareth Evans
 - Heather Thompson
 - Herb Sarnoff
 - Jonathan Rios
 - Kelly Carpenter
 - Laura Klesse
 - Michael Fisher
 - Miranda McManus
 - Nicole Ullrich
 - Noelle Larson
 - Renie Moss
 - Scott Plotkin
 - Vandana Akshintala
 - ...and more!

