The Diagnostic Odyssey
- Dr. Neena Nizar

www.thejansensfoundation.org
Patient Pathway to Cures/Treatment

www.thejansensfoundation.org
• A rare disease

• Miles and miles of desert sand

• No Google

• No “expert opinions”
Christmas, 2009

No Sign of Disease

Arshaan Adam  (A Strong and Brave Man)
Christmas, 2010

A Diagnosis at last

Jahan Adam (Savior)
Jansen Metaphyseal Chondrodysplasia (JMC)

• A rare disease of bone and mineral ion physiology
• ~ 30 patients known to date
• Autosomal dominant, caused by activating mutations in the PTH/PTHrP receptor (PTHR1)
• Short stature, skeletal abnormalities, hypercalcemia/hypercalciuria, nephrocalcinosis, renal disease.

www.thejansensfoundation.org
Dr. Harald Jueppner had been researching Jansen’s Disease for 20 years, but *never* met a patient.
The Jansen’s Foundation formed in February, 2017.

Jansen’s patient recovering from eardrum reconstruction, was born with craniosynostosis - the fusion of the skull's bones, and at two years old had to have his skull broken to make room for his brain to grow.
Jansen’s Foundation and MGH

Connected with the research team at MGH, and has been supporting the research efforts.
Children Living with Jansen’s

“During childhood, affected individuals may begin to exhibit progressive stiffening and swelling of many joints and/or an unusual ‘waddling gait’ and squatting stance.”

Post-Surgery Recovery

A few months post-surgery, the “Bends” are already back

Jansen’s patient’s legs pre-surgery

Picture Source: Jansen’s Foundation website:
See where our Jansen's Warriors Are At.
All 5 Jansen’s warriors in America

Summer 2017

www.thejansensfoundation.org
Creating Awareness for Jansen’s

Meet the Rare Family Who Account for 10% of All Cases of Jansen’s
There Are Just 30 Known Cases of Jansen’s Metaphyseal Chondrodysplasia Worldwide

Nebraska’s Neena Nizar Seeks Cure for Jansen’s, One of World’s Rarest Diseases

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Inverse agonist (IA) improves bone parameters of C1-HR mice
The Jansen’s Foundation Mission

The Jansen’s Foundations goal is to speed up research process, obtain all the necessary approvals, and to start the first testing of a disease-modifying peptide in an adult patient with Jansen’s disease.

- Nov. 2017 – RO1 NIH grant – pre-clinical studies
- June 2018 – Pre-IND meeting with FDA
- Sept 2018 – Patient Registry
- 2019 – NCATS TRND grant

2022 – FIH PROTOCOL TO FDA

www.thejansensfoundation.org
Never doubt that a small group of thoughtful, committed people can change the world.
Warriors of Hope!
Thank You!