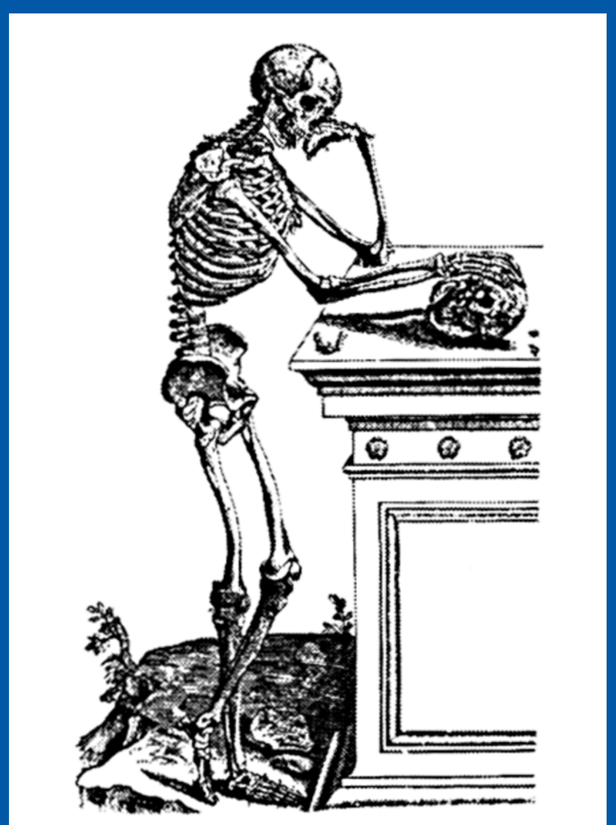
42nd Annual Meeting, Scottsdale, Arizona

Society of Skeletal Radiology

March 10-13, 2019

CASE OF THE DAY: MONDAY

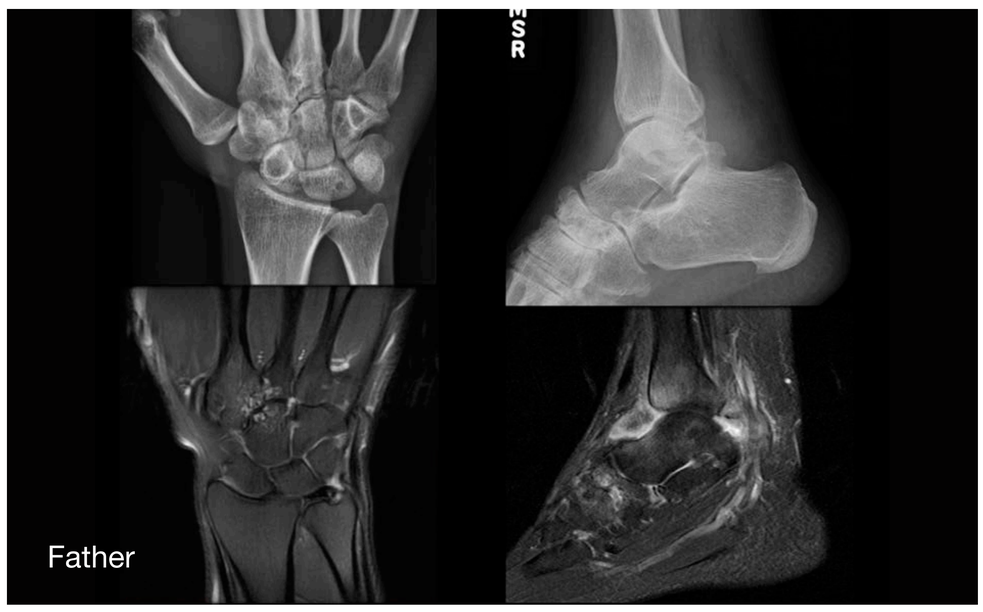
#SSRBONE19COD3



Kaushal Mehta, MD; Jeffrey Youngquist, MD; Ergent Zhiva, MD
University of Cincinnati, Cincinnati, OH

HISTORY

- 31-year-old father and 15-month-old son
- Father presents with 5 years of diffuse arthralgias and non-radiating back pain
- Son developed pain post-BCG vaccination



DIAGNOSIS

IFN-GR-1 Immunodeficiency with recurrent Atypical Mycobacteral infections in father and Disseminated BCG disease in the son

- IFN-GR-1 deficiency leads to impaired immunity and, consequently, severe and recurrent infections with Bacillus Calmette-Guerin (BCG) and atypical mycobacteria
- Autosomal recessive, Very rare (< 1/1,000,000)
- Caused by mutation on chromosome 6q23-q24
- Disseminated infection can involve bone, soft tissue, lungs, skin, lymph nodes
- Symptoms- Fever, weight loss, hepatosplenomegaly, lymphadenopathy, skin lesions
- Treatment- Hematopoietic stem cell transplant is the only curative Rx
- Prognosis- Poor in complete IFN-GR-1 deficiency with most patients not living past 10 years