Recurrent Necrotizing Pneumonia in a Patient with Langer-Giedion Syndrome

Marina A. Tipold DO/PGY-1, Samantha Wilson MD
Peyton Manning Children's Hospital – Ascension St. Vincent

Introduction

Langer-Giedion Syndrome (Trichorhinophalangeal Syndrome Type II) is a rare autosomal dominant genetic disorder commonly diagnosed following birth. This case report demonstrates that although Langer-Giedion Syndrome is rare, the recognition of this syndrome, the phenotypic spectrum, and potential complications are critical to diagnosis, research and development of new therapies, and possible prevention of disease progression.

Presentation

- Patient is a twelve-year-old male with known Langer-Giedion Syndrome.
  - PMH: multiple orthopedic surgeries, nocturnal gastrostomy tube feedings for improved nutrition, history of GERD status post Nissen fundoplication, sensorineural deafness status post cochlear implants, developmental delay, and coarctation of aorta status post repair with bicuspid aortic valve.
- Presented to ED with a four day history of fever, cough, and shortness of breath.
- CT with contrast performed: Large consolidation in the right upper lobe with central cavitation likely necrotizing pneumonia. Cystic lesions progressed from 2014.
  - This was the second occurrence of necrotizing pneumonia in this patient over a one year period.

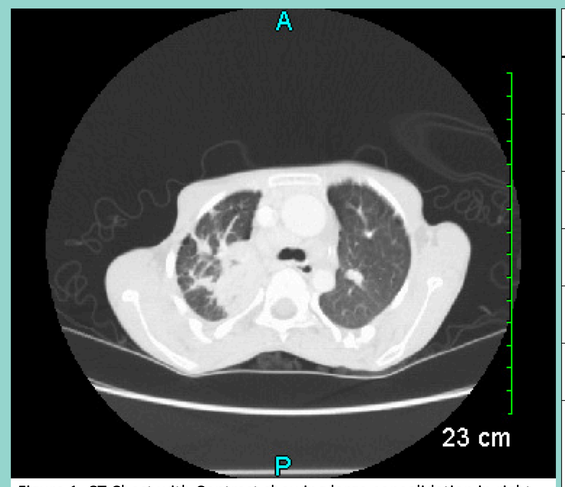

Figure 1: CT Chest with Contrast showing large consolidation in right upper lobe.

Workup:	
COVID PCR	*Negative*
COVID IgG	*Negative*
Fungal Panel	*Negative*
RP PCR	*Negative*
TB	*Negative*
Flu A + B	*Negative*
Group A Strep	*Negative*

Figure 2: Etiology Workup

Management

- Infectious disease consulted following diagnosis, and he was started on IV Clindamycin and Rocephin.
  - Following first antibiotic treatment in ED, patient started to become hypotensive and tachycardic with deterioration likely secondary to bacteriolysis from antibiotic treatment.
- He was switched to IV Vancomycin.
- Required oxygen supplementation and was monitored closely, with improving respiratory status over a five day period.
- Repeat CXR one week later, showed improvement.
- He was discharged home on three week course of oral clindamycin and cefdinir.
- Close follow up after discharge with Infectious Disease and subspecialists.

Discussion

- Langer- Giedion Syndrome
  - Deletion: portion of chromosome 8
  - Characteristics: short stature, dental manifestations, multiple bony exostoses, mental retardation, and characteristic facial features
  - Prevalence: estimated at 0.2-1/100,000 and likely underdiagnosed
- This patient continued to improve clinically following antibiotic course.
- Suspect aspiration of functional etiology, rather than immunologic. Basic immunodeficiency work-up was negative.
  - He had a history of poor growth trajectory, as commonly seen with the syndrome, but no history of dysphagia or aspiration.
- Pleural blebs noted on imaging, may be associated with connective tissue disorders related to genetic abnormality.
- There are currently no reports linking Langer-Giedion Syndrome and immunodeficiency or recurrent severe lower respiratory tract infections published in medical literature.
  - There are connections to recurrent ear and sinus infections, secondary to facial dysmorphisms.

Conclusion:

Further research is required to establish the connection and prevalence between Langer-Giedion Syndrome and recurrent pulmonary manifestations, specifically necrotizing pneumonia.

References:

1. Katge FA, Rusawat BD, Shivasharan PR, Patil DP. Langer-Giedion Syndrome: a Rare Case Report. *J Dent (Shiraz)*. 2016;17(3):238-241.
2. Konala P, Kiely N, Noakes C, Blair E, Cassar-Pullicino VN. Multiple long bone cysts revealed by MRI in trichorhinophalangeal syndrome type II predisposing to pathological fractures. Pediatr Radiol. 2017 Jul;47(8):1016-1021. doi: 10.1007/s00247-017-3839-4. Epub 2017 May 10. PMID: 28493010.
3. Lüdecke HJ, Wagner MJ, Nardmann J, La Pillo B, Parrish JE, Willems PJ, et al. Molecular dissection of a contiguous gene syndrome: localization of the genes involved in the Langer-Giedion syndrome. *Hum Mol Genet.* 1995; 4: 31–36.
4. Nagori SA, Jose A, Agarwal B, Bhatt K, Bhutia O, Roychoudhury A. Traumatic bone cyst of the mandible in Langer-Giedion syndrome: a case report. J Med Case Rep. 2014 Nov 25;8:387. doi: 10.1186/1752-1947-8-387. PMID: 25421062; PMCID: PMC4364690.