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Visual Case Discussion

Pediatric with Gaucher disease and Covid-19: Case report of uncommon manifestation of Covid-19 in chest Ct

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1. Discussion

The use of non-invasive diagnostic tools, fast and available with acceptable safety along with Rt-PCR is very important for better and faster management of COVID-19 patients. One of these diagnostic tools is the use of Non-enhancing Chest CT-scan.¹

The hallmarks of COVID-19 infection on imaging is bilateral and peripheral ground-glass and consolidative pulmonary opacities.² Moreover, opacities with a rounded morphology, reticulation, peripheral location of the opacities, consolidation with ground-glass opacities, and crazy-paving pattern had been reported.²

This is a rare case presentation of lysosomal storage disorder (Gaucher Disease- type 1) and sever Congenital Neutropenia (HAX1 mutaion -Kostmann Syndrome). Our patient is a rare report of the combination of Gaucher Disease and Kostmann Syndrome in the literature. Our patient does not have severe clinical presentation of infections due to severe neutropenia and hospitalization till now. Also in this admission she had just fever and cough without dyspnea. So it might be that association of Gaucher disease and enzyme replacement therapy may result as a modulating factor in clinical course of severe congenital neutropenia. Melis Demir Kose and et al. also reported a case such as our

patient with coexistene of Gaucher disease-type 3 and sever congenital neutropenia at the first year of life in 2019.³ Scientific research indicated that individuals with Gaucher's disease (GD) may be at increased risk of infection or complications of SARSCoV-2. There are lysosomal disruption in both GD and the COVID-19 moreover inflammatory responses play a key role in both diseases.⁴

Due to the wide spectrum of inflammatory reactions, cytokine secretion and myeloid cells involvement in GD and severe congenital neutropenia, if these persons infected with 2019 novel coronavirus it may be lead to atypical manifestations.

2. Conclusion

After the outbreak of COVID-19, it was determined that the disease has a series of specific symptoms. In this study, Cavitation in Chest CT-scan as atypical and rare manifestation in a patient who infected with COVID-19 was detected. Therefore, it is recommended that clinicians consider atypical evidence of patients with COVID-19 who have metabolic and immunodeficiency diseases.

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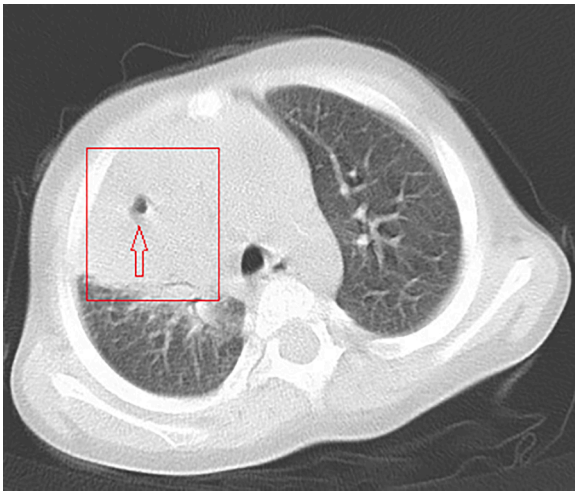


Fig. 1. Axial chest Ct without contrast demonstrate lobar consolidation in right upper lobe (Red box) with central cavitation change (Red arrow).

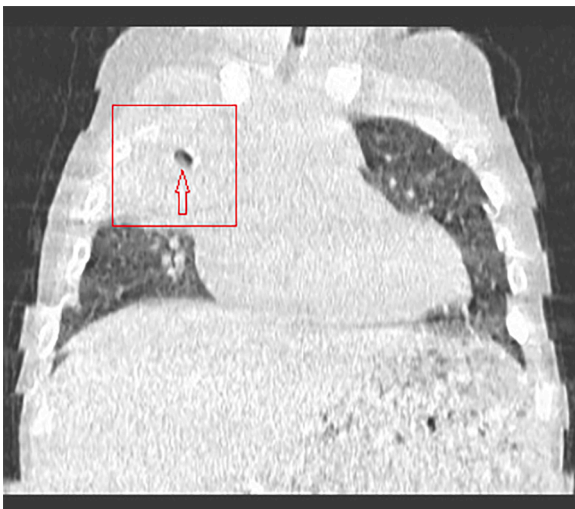


Fig. 2. Coronal chest Ct without contrast demonstrate lobar consolidation in right upper lobe (Red box) with central cavitation change (Red arrow).

3. Visual case discussion

A 2-year and 8-month-old girl known case of Gaucher disease type 1 and congenital neutropenia (GBA gene p.[Leu483pro];[Leu483pro], Exon11–12, Ch1, Unknown breakpoint and HAX1 c.661delG p.1222, Exon 5, Ch1: 154,247,734,Homozygote), that After diagnosis she received enzyme replacement therapy, was admitted with a chief complaint of fever and cough since 5 days before hospitalization in Mofid Children's Hospital, Tehran, Iran in April 27, 2020. She had a positive history of contact with a person infected with COVID-19 in the last month (the patient's parents).The management was done according to algorithmic approach to a child suspected to COVID-19 in Iran.⁵

Due to the history of contact with the infected person with COVID-19 and the suspicion to lower respiratory tract involvement, CBC, ESR, CRP, CXR and Covid-19 RT-PCR tests were requested for the patient. The results of laboratory data were as below:

WBC = $5.500 \times 10^3 / \mu\text{L}$ (Lymph : 77%, Neut : 15%), CRP = +++ ESR = 107mm/hr

CXR showed Right lobar consolidation, Also due to delay in preparing the Covid-19 RT-PCR test, Chest CT was requested for better

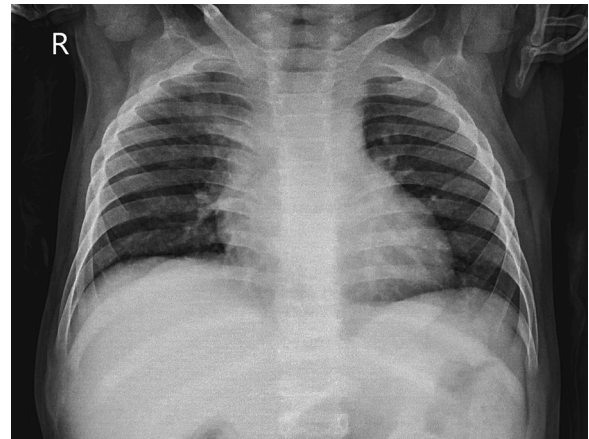


Fig. 3. Anteroposterior chest X-ray follow up shows complete resolution of the lesion in right upper lobe.

evaluation and better therapeutic decision. Chest CT findings included: Lobar consolidation in RUL with Central Cavitation change (Fig. 1 and 2). Imaging findings were atypical for COVID-19,² and bacterial pneumonia was included in the first differential diagnosis, Therefore, blood cultures were requested for the patient and empirical antibiotic therapy with ceftriaxone and vancomycin and also Granulocyte colony-stimulating factor (GCSF) was also administrated. .

After about 48 h, the result of PCR assay was positive for COVID-19, and also the blood culture test was negative, so Covid-19 treatment was started for the patient [hydroxychloroquine and azithromycin] .⁶ Patient was discharged after seven days from admission with resolution of clinical symptoms. One month after discharge, she was great clinically and CXR follow up showed complete resolution of the lesion (Fig. 3).

4. Questions and answers with a brief rationale

What can be the rare manifestations of Covid-19 disease in immunocompromised patients?

- Ground-Glass
- Consolidation
- Cavitation
- Crazy-Paving

Correct Answer = c

Conflict of interest

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Supplementary materials

Supplementary material associated with this article can be found, in the online version, at [doi:10.1016/j.visj.2021.100966](https://doi.org/10.1016/j.visj.2021.100966).

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