Qeios

Peer Review

Review of: "Two Cases of Non-Surgical Hypoparathyroidism That Were Diagnosed with Delay"

Mostafa Suhail Najim Najim[1]

1. Unity Health System, Rochester, United States

Thank you for sharing these interesting and challenging cases. It is clear that significant effort has gone into managing these patients. I do not have much comments on case 2 as it appears to be a work in progress, but here are my thoughts on case 1:

Mutations and Their Interaction:

If known or studied in the past, how do the mutations in LZTR1, TBX1, and CLCN1 potentially interact with or exacerbate each other in the context of the patient's clinical symptoms? Could you provide specific examples or studies that discuss the interaction of LZTR1, TBX1, and CLCN1 mutations?

Impact of Delayed Diagnosis or Treatment:

Was the delayed diagnosis or treatment a contributing factor to the persistence of symptoms? What alternative management strategies could have been considered if the diagnosis had been made earlier?

Diagnostic Confirmation of Neurofibromatosis Type II with Schwannomatosis:

Is the clinical presentation and the genetic mutations sufficient to establish a diagnosis of Neurofibromatosis Type II with schwannomatosis, or was there any additional imaging (e.g., MRI) or biopsy performed to confirm the diagnosis? This information would clarify the level of diagnostic certainty.

COVID-19 Impact:

The patient had multiple episodes of COVID-19 between 2020 and 2024. Is there concern that repeated viral infections, particularly COVID-19, may have worsened or exacerbated her underlying conditions, either directly or through immune system dysfunction?

doi.org/10.32388/GJUGKV

Psychiatric Symptoms and Management:

You mentioned a trial of psychiatric medications. Was there any indication that a psychiatric component contributed to her symptoms? The patient was also suggested to be treated in a psychiatric hospital in 2024. Could you elaborate on the rationale behind this recommendation? How did the patient respond to this suggestion, and were there any changes in her clinical course as a result?

Long-term Prognosis:

What is the long-term prognosis for this patient, given the complexity of her diagnoses? Understanding the expected course of her condition could help guide future management strategies.

Family History and Environmental Exposure:

How did the family history of exposure to the Chernobyl zone influence the diagnostic process and decision-making? Was radiation exposure considered a significant factor in her health issues, or was it ruled out early in the investigation?

Rationale for Active Vitamin D Treatment:

Although briefly mentioned in the discussion, could you elaborate on the physiological rationale for using active vitamin D (calcitriol) rather than ergocalciferol in this case? This is an important concept that would benefit the reader's understanding, particularly in cases of hypoparathyroidism or calcium metabolism disorders.

Declarations

Potential competing interests: No potential competing interests to declare.