



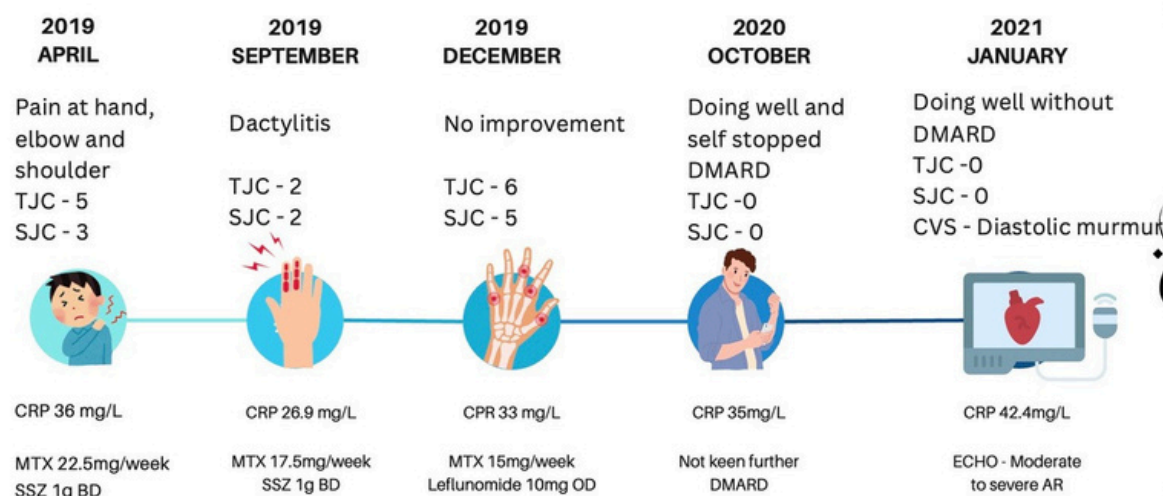
ID018: Primary Antibody Deficiency in a patient with inflammatory arthritis and porcelain aorta “A Tale of the Silent Guard and Ceramic Core”

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25 year old Man (2025)

Prolonged fever and pneumonia at 2 years old (2002)
Bilateral eye bphleritis at 10 years old (2010)
Otitis media with effusion at 11 years old (2011)
Peripheral SpA at 17 years old (2017) - Started Sulphasalazine

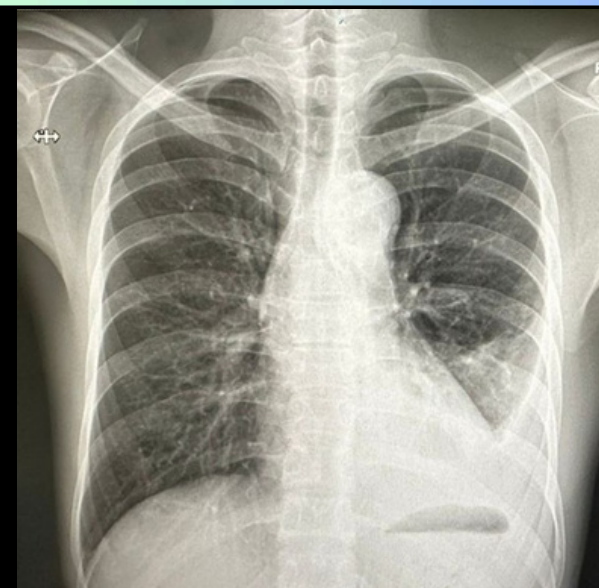
PROGRESS 2019 - 2021



Parapneumonic Effusion at 24 years old July (2024)

September 2024 - Lobar pneumonia and mitral valve infective endocarditis
Culture: Streptococcus Viridans (Sputum) & Haemophilus influenzae (Blood)

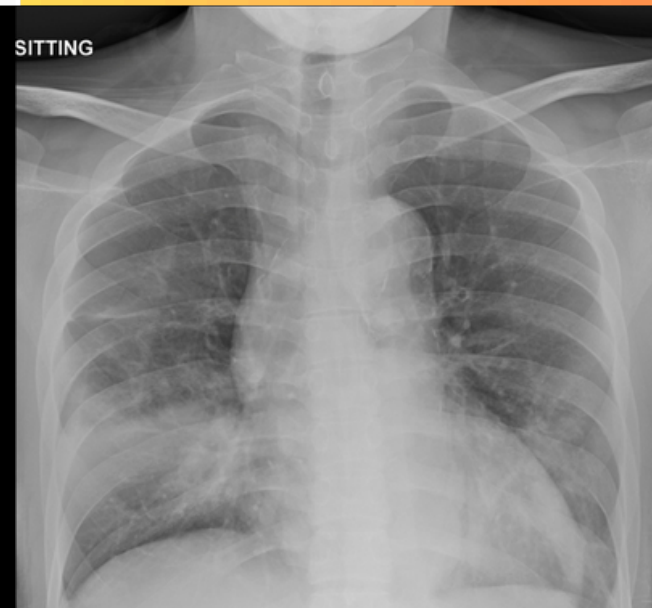
November 2024 - Lobar pneumonia
Proceeded with CT Scan and PET Scan



Chest Radiograph July 2024



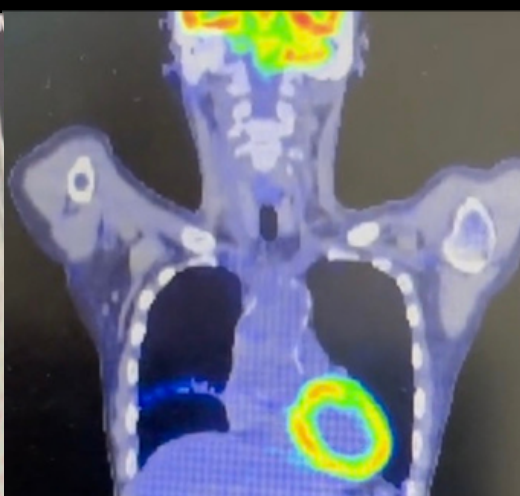
Chest Radiograph Sept 2024



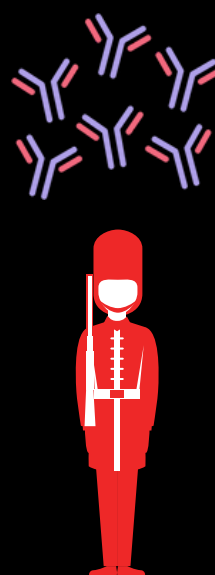
Chest Radiograph Nov 2024



CT November 2024



PET/CT November 2024



Discussion and Conclusion

- 1.Primary antibody deficiency associated arthritis shares features with SpA(Pott, RMD Open 2022)
2. To consider immunodeficiency in patients with recurrent infection
3. To consider differential of primary immunodeficiency in patients with persistent low globulin
4. Differential diagnosis of enlarged aorta include hereditary cause, inflammatory cause and infectious cause
5. PA (Porcelain Aorta) is associated with systemic inflammatory diseases such as Takayasu arteritis (TA) and need to be considered in this patient (Abramowitz et al. Circulation. 2015)
6. There are rare reported case of Takayasu arteritis with primary immunodeficiency (Sener et al, 2022)

Investigations

Hemoglobin 13.7g/dL
Sodium 140 mmol/L
Creatinine 48 umol/L
CRP 64 mg/L (high)
HIV Serology Negative
Syphilis Negative
Albumin 32 g/L ,
Globulin 16g/L (low)
IgG < 3 (low), IgA 0.1 (low),
IgM <0.04 (low)

-Elder brother died due to infection at age 26
-Male cousin passed away due to infection at age 29
-2nd male cousin has bronchiectasis with recurrent infection on monthly hospital visit

Gene Sequence Analysis: BTK Gene PATHOGENIC
Flow cytometry assay -
66% patient monocyte express BTK Protein

DIAGNOSIS

X-Linked Agammaglobulinaemia (Brutons) with Primary Antibody Deficiency (PAD) associated arthritis and Porcelain aorta