A culture-negative mycobacterium infection in a boy with familial primary immunodeficiency disease

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INTRODUCTION:

Primary immunodeficiency diseases (PIDs) are rare inherited disorders that impair the immune response leading to increased risk of infections, autoimmune phenomena, and malignancy.[1,2] We report a case of culture negative mycobacterium infection of distal radius of a child with familial PID.

REPORT:

10 years old boy who a right-hand dominant presented with left wrist swelling and pain which gradually worsening for 2 months prior to presentation. No significant constitutional symptoms. There was 5x5 cm bony hard swelling over lateral aspect left distal forearm, with warm and tenderness. Otherwise, he was generally well.

He was the eldest from 6 siblings. He had NICU admission at day 10 of life for sepsis which resolved with antibiotics. He had episodes of generalised cutaneous rashes which was self-remitting. Two of his brothers were diagnosed with PID with disseminated tuberculosis (TB) at neonatal period as presented with generalised lymphadenopathy. Despite completion of anti-TB medication, one of them died from brainstem encephalitis at 4 years old.

Our patient was investigated for PID and TB infection. ESR was elevated, however total white and differential count were normal. Tuberculin skin test (TST) was significant. Interferon-gamma release assays for detection of TB and PID screening were pending. Plain radiograph of radius/ulna showed osteolytic lesion at metaphysis of distal radius with honeycomb appearance. MRI of forearm depicted bone changes of distal end radius with cortical and epiphyseal plate erosion, periosteal reaction, phlegmonous soft tissue enhancement that suggestive of chronic infective bone changes with possibility of TB. Findings from an open biopsy revealed multiple sinuses from the bone, present of intramedullary slimy and whitish slough with thick periosteum. Histopathology result showed acute on chronic inflammatory process, however acid-fast bacilli and mycobacterium were not detected. Bone culture and gene expert for TB and fungal infection were negative.

Despite negative culture result for TB, we treated him with anti-TB based on positive TST and strong family history of TB infection. He responded well with treatment and serial follow-up plain radiograph depicted sign of recovery.

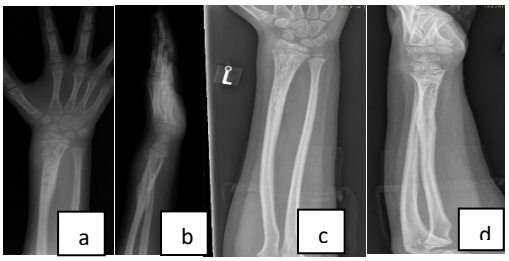

Figure 1: Plain radiograph of radius/ulna showed osteolytic lesion at metaphysis of distal radius with honeycomb appearance (a,b). After 6 months of anti-TB, previous lytic lesion being filled by new bone (c,d).

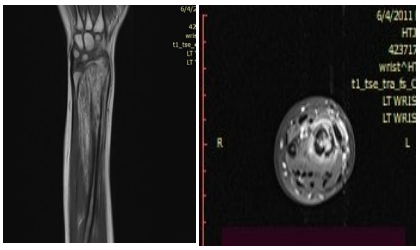

Figure 2: MRI coronal and axial view of left radius ulna

CONCLUSION:

TB remains the common cause of chronic insidious osteomyelitis despite a negative culture especially in immunocompromised patient. Clinical history and radiographs findings were excellent tools to guide our management to prevent devastating TB complication.

REFERENCES:

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