

TEACHING FILES (GRAND ROUNDS)

FAMILIAL TUBERCULAR ABSCESSSES

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Clinical Problem

A 13-month-old boy presented with an abscess over the right thigh following measles vaccine. The abscess was drained and histopathology of the abscess wall showed non-caseating granulomatous inflammation suggestive of atypical mycobacterial infection. Cultures and PCR tests were not done. His Mantoux test was positive (12 mm) and chest X-ray was normal. Paternal cousin brother had axillary BCG adenitis requiring incision and drainage along with anti-tubercular therapy (ATT) for six months. His mother also had tuberculous abscess over the back 9 years ago and required incision and drainage along with ATT. This child was started with isoniazid, rifampicin, clarithromycin, ethambutol, and ofloxacin. ATT was stopped after six months.

Does this child have a genetic predisposition to tuberculous infection?

Discussion:

Abscesses due to injections are generally iatrogenic infections caused by improper aseptic measures or contaminated needles. Pyogenic organisms like *Escherichia coli*, *Klebsiella spp*, *Staphylococcus aureus* are the common agents causing these abscesses.¹ However, rapidly growing non-tuberculous mycobacteria (NTM) like *Mycobacterium chelonae*, *M. fortuitum* or even *M. tuberculosis* can rarely cause sterile caseating abscesses at parenteral injection sites.¹ Most commonly NTM infection is because of contamination with tap water as that is the reservoir of these organisms. It is important for the clinician to know mycobacteria as a probable cause of injection abscesses especially when the aspirate is sterile so that proper anti-tubercular therapy (ATT) or in cases of NTM, treatment with antibiotics after susceptibility testing (against amikacin, doxycycline, fluoroquinolones, clarithromycin along with first-line antitubercular drugs) can be commenced.¹

Development of abscesses after infection with mycobacteria is dependent on multiple host and pathogenic factors. The susceptibility of the host to

develop abscesses or even tuberculosis (TB) is modified by genetic factors (which is one of the reasons why only about 10% of infected individuals eventually develop pulmonary TB).² Hence certain individuals have a genetic predisposition to develop mycobacterial abscesses. Various genetic polymorphisms including chemokine (C-C motif) ligand -2 (CCL-2), interleukin-8 (IL-8), toll-like receptor (TLR), natural resistance associated macrophage protein-1 (NRAMP-1), nucleotide-binding oligomerization domain-containing protein 2 (NOD 2) have been implicated to cause susceptibility to mycobacteria.³⁻⁸ Most of these genes participate in the immune response and hence these polymorphisms alter the immune system and make the individual susceptible to mycobacterial disease. Another rare congenital syndrome responsible for enhancing the susceptibility to mycobacteria is Mendelian susceptibility to mycobacterial disease (MSMD).⁹ Mutations in genes encoding any of the molecules involved in the interleukin 12 (IL-12)/ interferon-gamma (IFN- γ) axis could be responsible.⁹ These genetic polymorphisms may be passed down to the patient's offspring and may be responsible for a family history of mycobacterial infections sometimes in the form of injection abscesses as seen in the above case. Molecular diagnosis will have to be carried out to rule out the major genetic alterations.¹⁰

Compliance with ethical standards

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