

## **Imaging inflammation in early-onset sarcoidosis: granulomatous anterior uveitis in a 7-year-old girl**

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A 7-year-old girl, with early onset sarcoidosis, attended our clinic for a routine follow-up eye examination. She had reported no recent visual problems.

The patient had been diagnosed with sarcoidosis aged 4 years after a 6 month worsening history of problems walking, rashes on her face and upper limbs associated with fever, and red eyes. Arthritis and uveitis were confirmed on admission, alongside elevated ACE (436 U/L) and ESR (52mm/hr).

Chest X-ray was normal. Synovial biopsy revealed non-necrotising granuloma, and genetic testing revealed a novel mutation of unknown significance in the NOD2 gene. Her inflammatory changes had improved with treatment with methotrexate (weekly) and adalimumab (fortnightly) both given subcutaneously by parents at home. On examination during routine review, we found that she had reduced visual acuity in both eyes: 20/40 in the right and 20/60 in the left (typical 20/20).

A slit-lamp examination showed bilateral anterior chamber inflammation and large off-white deposits on the inner corneal surface: known as mutton-fat keratic precipitates. Anterior-segment optical coherence tomography showed inflammatory cells in both eyes and the keratic precipitates (figure). This recurrence of uveitis was associated with raised ESR (41mm/hr).

We started the patient on topical corticosteroids: 0.1% dexamethasone eye drops TDS for one week then reduced to BD, and also supported the family with systemic therapy concordance.

At follow up four weeks later, both the patient's vision and the anterior chamber inflammation had improved. The patient's concordance with methotrexate and adalimumab had also improved: previously the family had omitted doses having assumed that the long period of disease inactivity was due to remission rather than disease control.

Sarcoidosis is a rare, multisystem chronic inflammatory disorder, most commonly affecting the lungs (adult onset disease) and skin (childhood sarcoid). Early onset disease, which starts before the age of 4 years, typically presents with the triad of skin, joint and eye involvement, and may be caused by mutations in the NOD2 gene. Eye signs in early and later childhood onset include anterior uveitis, vitritis, choroiditis and retinal vasculitis.

Anterior uveitis may be asymptomatic in children up until the development of irreversible sight-threatening complications. Although anterior chamber inflammation is often difficult to detect, mutton-fat keratic precipitates can be seen with the naked eye. The presence of these large deposits—aggregates of polymorphonuclear cells, lymphocytes, and epithelioid cells—suggests granulomatous disease indicating differential diagnoses and investigations.

Clinical features are key to identifying the underlying granulomatous disorder when mutton-fat keratic precipitates are seen. Prompt diagnosis and management of the associated systemic disease, with coordinated care involving specialists, affords the best opportunity of disease control, and reduced burden of avoidable later-life morbidity and disability.

### **Contributors**

We all provided care for the patient. We have all verified the data and approved the final draft. ALS conceptualised the article. Written consent for publication was obtained from the patient's mother

**Declaration of interests**

We declare no competing interests.

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**Figure: Granulomatous anterior uveitis in early-onset sarcoidosis**

Anterior-segment optical coherence tomography showed inflammatory cells (arrowheads) in the right and left eye (A and B respectively) along with the keratic precipitates (dotted arrows)

**Multiple-choice question**

Which is NOT a most common morbidity in early-onset sarcoidosis?

- A) skin rashes
- B) polyarthritis
- C) recurrent uveitis
- D) visible pulmonary involvement

Answer D)

Using images (A) and (B)