SESSION IV

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Identical twins, one presenting as tuberculosis, the other as sarcoidosis

CASE PRESENTATION

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Case 1

A 21-year-old Irish factory worker who had been in England for 4 years presented with a 2-week history of a febrile illness with rigors, weight loss, productive cough and lassitude.

On admission to hospital he was found to have a right-sided pleural effusion without evidence of a parenchymal lesion. Pleural aspiration and biopsy were performed. No tubercle bacilli were demonstrated in the pleural fluid, either by direct examination or on culture. The histology of the pleural biopsy showed granulomatous pleurisy with caseating epithelioid and giant cell granulomata characteristic of active tuberculosis (Fig. 1). The Mantoux test (10 t.u. PPD Weybridge) was positive (20 mm induration) when read at 72 hours. He was treated with streptomycin 1 g, sodium PAS 12 g and isoniazid 300 mg daily for 3 months and sodium PAS and isoniazid for a total period of 18 months. Steroids were given initially for 4 weeks. The chest radiograph showed complete clearing of the effusion and no parenchymal lesion was subsequently demonstrated. A Kveim test (Spleen K.12) was performed 18 months later. The Kveim test site was biopsied at 5 weeks and the histology showed multiple discrete epithelioid and giant cell granulomata in the deeper dermis (Fig. 2) with inclusion bodies in some of the giant cells. A further Kveim test (Hurley) 2 months later showed no evidence of sarcoid granulomata on serial section. Subsequent follow up at 2 years showed him to be well, working and with a normal chest radiograph.

FIG. 1 (upper). Pleural biopsy. Note epithelioid and giant cell granulomata with caseation.

FIG. 2 (lower). K.12. Multiple semi-confluent granulomas.

FIG. 3 (upper). Mediastinal lymph node with characteristic granulomas. No suggestion of caseation.

FIG. 4 (middle). K.12. Peripheral satellite granulomas adjacent an area of fibrinoid necrosis.

FIG. 5 (lower). Hurley. More loosely arranged and widely scattered granulomas, but nevertheless a positive reaction.

Case 2

The twin brother came to England at the same time and they lived in the same household. He was examined as a contact 2 months after his brother was diagnosed as suffering from the tuberculous pleural effusion. He was well and symptom-free, but his chest radiograph showed bilateral hilar gland enlargement. The Mantoux test (10 t.u. PPD Weybridge) was positive (15 mm induration) when read at 72 hours. Serum proteins, electrophoretic strip, alkaline phosphatase, serum and urinary calcium estimations were normal. No bone cysts were present. The lachrymal and salivary secretions were normal. There was no evidence of ocular sarcoidosis. Mediastinoscopy was performed; the histology showed epithelioid cell and giant cell granulomata becoming confluent and with extensive hyaline fibrosis (Fig. 3). No tubercle bacilli were demonstrated by direct examination, culture or guinea pig inoculation. The bronchial mucosa was normal and histologically free from sarcoid granulomata. The Kveim test (K.12) showed a 3 mm papule after 5 weeks and serial sections of the Kveim test biopsy showed typical sarcoid granulomata (Fig. 4). A subsequent Kveim test (Hurley) 2 months later was also positive (Fig. 5). His tuberculin test (10 t.u. PPD) was repeated 18 months later when his chest radiograph showed little change; it remained positive (18 mm induration). Two years later he remained well. Although no treatment was given at any time, his chest radiograph showed some regression of the bilateral hilar gland enlargement.

Both twins were investigated to establish the genetic evidence for their identity. These results were as follows:

O; CDe/CDe; Ms/Ns; P₁+; Lu(a-); K-:Fy(a+)
 Hp:2-2; Gm(a+x-q+g+b+); Tf:C;
 AP:CA; PGM:1; Ak:1; Pep A:1; Pep B:1;
 Pep C:1; PHI:1; Ada 1.

These investigations show that the twins have the identical genotype-phenotype pattern and the probability of two non-identical twins producing this is less than 1 in 50. They are, therefore, in all likelihood, identical twins.