Eosinophilic Fasciitis and related disorders in families

From the published literature

(1) Eosinophilic Fasciitis in a pair of siblings (1989).1

"Two siblings, a 38-year-old woman and a 33-year-old man, developed eosinophilic fasciitis within a period of 6 months. They were found to have identical HLA-A, B, DR, and DQ antigens, raising the possibility of a genetic influence in the development of this disease. No common environmental factors close to the time of onset were identified; however, the possibility of a common, remote environmental factor cannot be discounted."

(2) Eosinophilic Fasciitis in a father and son (1994).²

"HLA typing was performed on the father and son, which showed the son inherited A3, B52, Cw null haplotype from his father. The two patients described appear to be the first cases of eosinophilic fasciitis involving a father and son. . . The only other familial cases of eosinophilic fasciitis is the report of siblings with identical HLA genes. Our patients, however, had completely different haplotypes from those described."

"We feel eosinophilic-fasciitis probably has a multifactorial aetiology with a combination of genetic and environmental factors playing a role."

(3) Familial eosinophilic fasciitis and breast cancer (1994).3

"Eosinophilic fasciitis (EF) is an uncommon condition which unlike systemic sclerosis has only rarely been associated with solid malignancies. We report the occurrence of EF in **two sisters**, both of whom developed contemporaneous breast cancer."

(4) Eosinophilic Fasciitis in Siblings (2013).4

"We describe a case of siblings with EF and identical HLA types, suggesting a genetic predisposition. The reporting of unique cases such as these is important in furthering our knowledge of rare conditions."

"Patient 1 presented at 14 years of age with a 3-month history of upper limb joint stiffness and swelling after prolonged physical exertion with associated general malaise and weight loss... A biopsy demonstrated a diffuse inflammatory cell infiltrate in the fascia, with eosinophils, lymphocytes, and plasma cells. She responded well to prednisolone and is maintained on oral methotrexate."

"Patient 2, the younger brother and only sibling of Patient 1, also presented at age 14 years with a 3-week history of skin thickening on his upper limbs, with venous guttering and digital sparing... A clinical diagnosis of EF was made and he responded to oral prednisolone and methotrexate."

"EF remains poorly understood. This report suggests that family members with similar HLA typing are at increased risk, with a possible link to HLA-A2. It is likely that a second environmental factor contributes to the timing of disease onset. The low disease prevalence makes it difficult to do the case-control studies needed to confirm a correlation with HLA."

(5) Familial eosinophilic fasciitis induced by toxic oil (1988).5

"Three of four family members in husband, wife and one of two daughters developed in a short time interval (eleven months) eosinophilic fasciitis. The clinical, analytical and histopathological changes were characteristic of this disease. Familial cases of eosinophilic fasciitis have not been previously published. The process of these patients was probably caused by the ingestion of denatured oil (toxic oil syndrome), although the clinical picture begun two and a half years after the epidemic phase of the toxic oil syndrome declined."

(6) Eosinophilic fasciitis in cousins (2011).6

"Family history was significant for a **first cousin** with EF . . . our case is now the third familial case reported in the United States² and Canada,¹ suggesting that family history is likely to be a risk factor."

From online patients forums

(7) Eosinophilic Fasciitis in twin sisters.

The first sister developed EF in 1996 aged 10, the second six years later aged 16.

(8) Eosinophilic Fasciitis and Eosinophilic Esophagitis in the same family.

The wife of an EF patient posted that her husband had been diagnosed with Eosinophilic Fasciitis and Eosinophilic Esophagitis. His father had Eosinophilic Asthma and his two sons had Eosinophilic Esophagitis.

(9) Eosinophilic Fasciitis and Eosinophilic Myositis in the same family.

An adult female with Eosinophilic Fasciitis posted that her father and his brother both had Eosinophilic Fasciitis. Her father also had Eosinophilic Myositis.

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