Chronic granulomatous disease andlupus erythematosus

Introduction: Chronic granulomatous disease (CGD) is a rare hereditary primary immunode?ciency with several forms, most commonly of X-linked inheritance. In addition to increased susceptibility to bacterial and fungal infection, patients with CGD are also predisposed to a variety of in?ammatory and autoimmune conditions, including systemic lupus erythematosus (SLE). There have also been reports of discoid lupus, photosensitivity and other lupus spectrum dermatoses, especially among female carriers of X-linked CGD. The pathogenesis of such dermatoses in patients and carriers of CGD is poorly described.
Objective: To describe two cases of lupus spectrum dermatosis in boys with CGD and hypothesise on pathogenenic pathways.

Case details: One boy with CGD developed intermittent, photosensitive facial erythema from the age of 6 months. At age 7, he developed generalised erythematous plaques, histologically consistent with lupus erythematosus. He also had recurrent mouth ulcers, lip swelling and anal ?ssures. Serial dihydrorhodamine oxidation (DHR) test performed over several years revealed that his proportion of functional neutrophils ?uctuated between 23% and 90%. A second case is presented which also shows partial neutrophil oxidative function in association with cutaneous lupus.

Conclusion: The link between CGD and lupus-spectrum dermatoses remains unclear. Abnormal apoptosis and NETosis have both been implicated in lupus pathogenesis. We hypothesise that having a proportion of functional neutrophils, with exaggerated NETosis associated with abnormal apoptotic stimuli, may play a role in the pathogenesis of lupus in some CGD patients and carriers.