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The autoimmune and immunodeficiency manifestations of ACP5 mutations

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Spondyloenchondrodysplasia is a rare immuno-osseous dysplasia caused by biallelic mutations in the gene *ACP5*, which encodes tartrate resistant acid phosphatase (TRAP). I will present data pertaining to the recognized skeletal, neurological and immune phenotypes, most particularly the immune manifestations. In a recent analysis of 26 patients, 22 manifested clinical autoimmune disease, most frequently autoimmune thrombocytopenia and systemic lupus erythematosus and further two demonstrated positive autoantibodies. In the majority of patients tested we detected up-regulated expression of interferon-stimulated genes (ISGs), in keeping with the autoimmune phenotype and the likely immune-regulatory function of the deficient protein TRAP. Two mutation positive patients did not demonstrate an up-regulation of ISGs, including one patient with significant autoimmune disease controlled by immunosuppressive therapy perhaps demonstrating a useful treatment for the autoimmune manifestations. Of further note recurrent bacterial and viral infections were reported in five of 26 patients, raising the suggestion that immunodeficiency is a part of *ACP5*-associated disease. Interpretation of immunological testing undertaken in the cohort was difficult, in terms of differentiating disease-related immunodeficiency from immune defects resulting from immunosuppressive therapy. Whilst additional data are needed, we would recommend in the interim that patients with biallelic *ACP5* mutations should be monitored for an infectious susceptibility and should undergo lymphocyte phenotyping and serum immunoglobulin values prior to immunosuppressive therapy.

Biography

Tracy Briggs was qualified from Liverpool Medical School in 2003 and trained in Pediatrics and then Clinical Genetics. She undertook a PhD during her clinical training at The University of Manchester and is currently an NIHR Clinical Lecturer. She spends 50% of the time working in the Genomic Medicine Department at the Central Manchester NHS Foundation Trust and 50% of the time at The University of Manchester. Her research interest is immunogenetics, particularly innate and autoimmune genetic disorders.

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