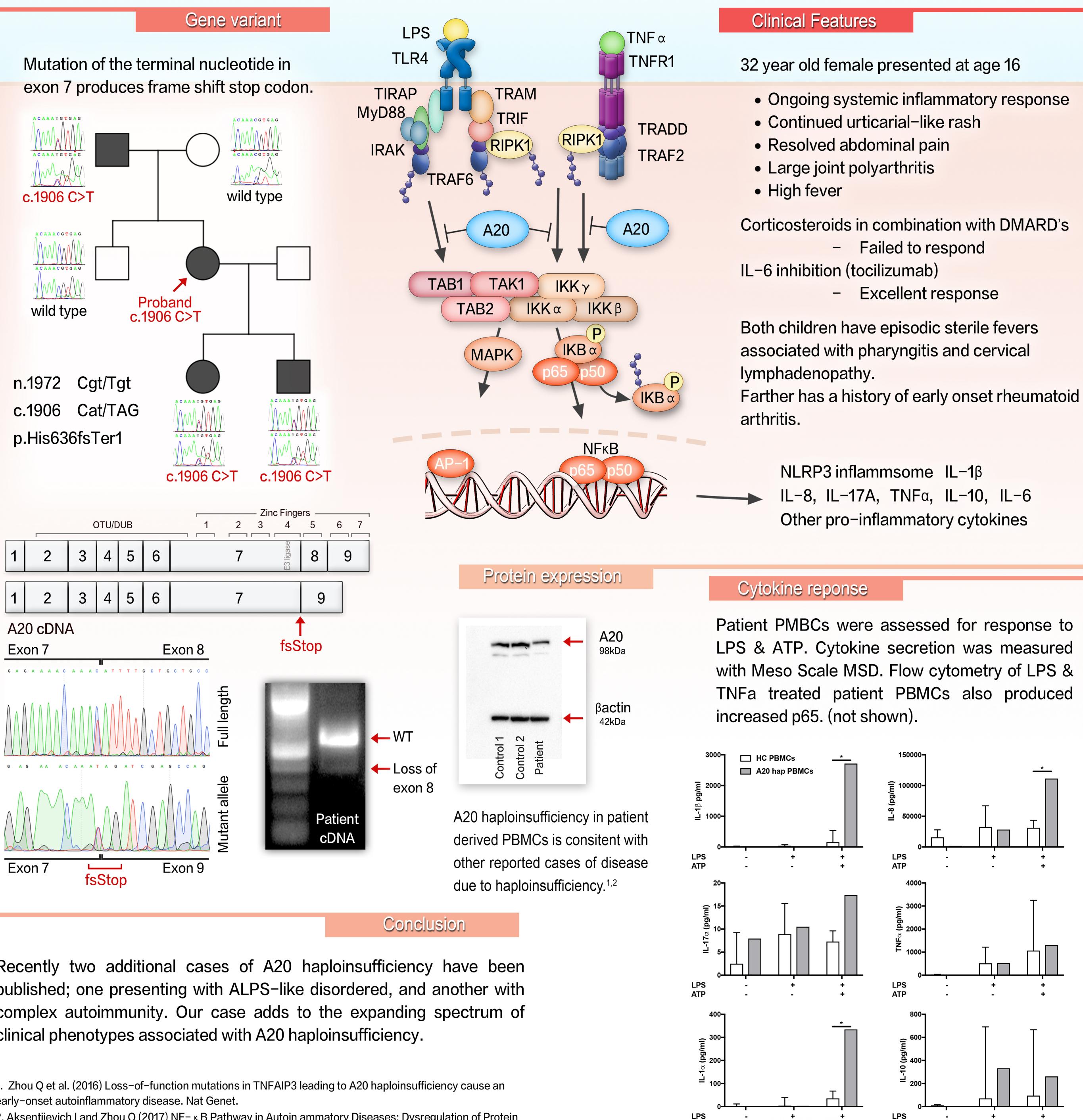


A case of Adult onset Still's Disease (AOSD) caused by a novel splicing mutation in *TNFAIP3* successfully treated with tocilizumab

Dylan Lawless¹, Thomas Scambler², Shelly Pathak², Laura Rice¹, Rashida Anwar¹, Sinisa Savic^{2,3,*}

TNFAIP3 encodes the NF-κB regulatory protein A20. High-penetrance heterozygous germline mutations in *TNFAIP3* have been recently described to cause a novel systemic autoinflammatory disorder due to haploinsufficiency of A20 and inadequate inhibition of NF-κB pathway.^{1,2} The majority of patients have presented with Behcet's disease-like phenotype characterised by recurrent oro-genital ulcers, uveitis and arthralgia. Here we describe a patient who presented with AOSD but found to have a novel splicing mutation in *TNFAIP3* causing A20 haploinsufficiency



1. Zhou Q et al. (2016) Loss-of-function mutations in *TNFAIP3* leading to A20 haploinsufficiency cause an early-onset autoinflammatory disease. *Nat Genet*.

2. Aksentijevich I and Zhou Q (2017) NF-κB Pathway in Autoinflammatory Diseases: Dysregulation of Protein Modifications by Ubiquitin De ned a New Category of Autoinflammatory Diseases. *Front Immunol*.

*Sinisa Savic MD, PhD Email: s.savic@leeds.ac.uk

¹Leeds Institute of Biomedical and Clinical Sciences, University of Leeds, Wellcome Trust Brenner Building, St James's University Hospital, Beckett Street, Leeds, UK.

²Leeds Institute of Rheumatic and Musculoskeletal Medicine, Wellcome Trust Brenner Building, St. James's University Hospital, Beckett Street, Leeds, United Kingdom.

³Department of Clinical Immunology and Allergy, St. James's University Hospital, Leeds, United Kingdom.



NIHR
Leeds Musculoskeletal
Biomedical Research Unit

UNIVERSITY OF LEEDS
The Leeds Teaching Hospitals NHS Trust

