Response to: 'A20 haploinsufficiency (HA20): clinical phenotypes and disease course of patients with a newly recognised NF- κ B-mediated autoinflammatory disease' by Aeschlimann *et al*

We thank Dr Berteau *et al*¹ for sharing their experience of a family diagnosed with HA20 based on a Behçet-like disease phenotype and an autosomal-dominant inheritance pattern.

The presentation and disease course described in the mother and her two children support the findings described in our cohort² and in a recent large cohort of Japanese patients with HA20.³ An interesting observation presented by Dr Berteau *et al* is the development of high fever and a severe local inflammatory swelling following immunisation with an unconjugated anti-pneumococcal vaccine in the mother. While we did not observe such reactions in our cohort, they have also been reported in several Japanese patients with HA20³ and in other autoinflammatory diseases such as the cryopyrin-associated periodic fever syndrome.

This letter reinforces our observations and adds a new mutation c.[994G>T] p.Glu332* to the list of already known heterozygous *TNFAIP3* mutations. In addition, it again highlights the need for clinicians to suspect HA20 in patients with Behçet-like disease phenotypes and the necessity for more research into this disease, from both clinical and basic research perspective. Of note, the patients included in our study were not all Americans, but Caucasians of Turkish, European, American and Dutch descent.

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Contributors RML and FAA drafted, revised and approved the response to the eLetter. The co-authors of the initial publication 'A20 haploinsufficiency (HA20): clinical phenotypes and disease course of patients with a newly recognised NF- κ B-mediated autoinflammatory disease' by Aeschlimann *et al* (ARD 2018) all approved the submitted response to the eLetter.

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