PEDIATRIC RHEUMATOLOGY

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P02-033 - CAPS diagnosis and treatment in an Israeli family

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Introduction

Only one family in Israel, from Ethiopian Jewish origin has been diagnosed with the familial cold autoinflammatory syndrome phenotype of the cryopyrin associated periodic syndromes (CAPS)[1].

Case Report

We confirmed the Muckle-Wells syndrome phenotype of CAPS by *NLRP3* genetic testing in a three generation family of Turkish Jewish origin, previously diagnosed with familial Behcet disease due to the presence of mucosal ulcers in several family members with the finding of the HLA-B51 antigen in at least one family member. Eight family members including a deceased grandfather, 4 of his daughters and three grandchildren had brief episodes of fever and chills, accompanied by headache, myalgia, arthralgia, and an urticarial skin rash. Most family members had substantial hearing loss. None developed amyloidosis. Four family members tested for a *NLRP3* pathogenic variant had the known NM_001243313.1: c.1043C>T, p.Thr348Met variant[2]. Following initiation of treatment with canakinumab (150 mg every 8 weeks) and colchicine for mucosal ulcers all disease symptoms resolved and acute phase reactants normalized except for persistent headaches in one grandchild and tinitus in another. The health-related quality of life of the treated grandchildren markedly improved.

Discussion

NLRP3 genetic testing was instrumental in the diagnosis of CAPS in this family, particularly as some family members presented with atypical features suggestive of Behcet disease, which is much more common in Israel. Although CAPS is a rare disease, additional cases with other *NLRP3* variants may exist in Israel.

Disclosure of interest

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References

1. Shalev SA, Sprecher E, Indelman M, Hujirat Y, Bergman R, Rottem M: **A novel missense mutation in CIAS1 encoding the pyrin-like protein, cryopyrin, causes familial cold autoinflammatory syndrome in a family of ethiopian origin.** *Int Arch Allergy Immunol* 2007, **143**:190-3.
2. Dodé C, Le Dû N, Cuisset L, *et al*: **New mutations of CIAS1 that are responsible for Muckle-Wells syndrome and familial cold urticaria: a novel mutation underlies both syndromes.** *Am J Hum Genet* 2002, **70**:1498-506.

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