Objectives: To describe the clinical characteristics and genetic variants of patients diagnosed with TRAPS syndrome in a cohort of patients with autoinflammatory syndromes with follow-up in a tertiary hospital from 2013 to the present.

Methods: Retrospective descriptive study of adult patients diagnosed with autoinflammatory syndrome since 2013 (year of introduction of genetic tests in the hospital laboratory) until now. The data was obtained from the review of medical records. All patients with mutations in TNFRSF1A gene and clinically compatible with this diagnosis were reviewed.

Results: Of a total of 44 adult patients diagnosed with hereditary syndromes of periodic fever (FMF, TRAPS, cryopyrinopathies, HIDS) and compatible genetic mutations (excluding polymorphisms), 13 (29.5%) presented mutations in TNFRSF1A gene. Of those 13 patients, 9 (69.2%) were women. The most frequent mutations were the mutation in TNFRSF1A gene in exon 4 (p.R92Q) with 12 cases (92.3%) and one case (7.6%) with mutation in heterozygosis in exon 3 (p.P46L). The mean age at diagnosis was 27.92 years (IR 12-55 years). 5 patients showed simultaneous genetic variants in other genes related to autoinflammatory syndromes. Signs and/or symptoms were myalgia and elevation of acute-phase reactants with 12 patients (86%) followed by fever and joint symptoms (arthritis and/or arthralgia) with 11 patients (79%) (Table 1). The most frequent 2 patients (14%) had cutaneous involvement. 2 cases (14%) presented lymphadenopathy. Regarding treatment, 5 patients (36%) had received treatment with biological therapy (anti-IL-1 or anti-TNF) and another 5 patients (36%) had received treatment with corticoids. The corticoids were used in 6 patients (43%).

Conclusion: The TRAPS syndrome is a clinical entity to consider when making a differential diagnosis in patients with suspected autoinflammatory syndrome, that present fever, acute phase reactants elevation, arthromyalgia and its confirmation diagnosis is with genetic test.

Disclosure of Interests: None declared