(VCSS) were used for the assessment of venous disease. Venous disease-specific QoL was measured through Venous Insufficiency Epidemiological and Economic Study Quality of Life/Symptom (VEINES-QoL/Sym) questionnaire.All patients were reanalyzed using color Doppler USG in the Radiology Department,by a radiologist.In each patient, a total of 16 superficial and deep veins in both legs were assessed for the presence or absence of obstruction, recanalization,reflux, and collaterals within 1 weeks following the clinical examination.

Results: During venous assessment,median disease duration was 9(0-34) years. Eighty(84.2%) patients were under ımmunsupressive(IS) treatment and 13 of these patients were under anticoagulation treatment in addition to ISs. Duration between first vascular event and venous assessment was 6(1-26) years. PTS was present in 57(61.3%) out of 93 patients and severe PTS was present in 19(19.8%)patients.There was no association between the presence of PTS and sex,disease diagnosis age,age during DVT,presence of relaps. There was no difference between patients with or without PTS according to the anticoagulant usage (p=0.817).Doppler ultrasound examination showed bilateral at 31(31.4%) patients and both upper and lower involvment at 40(47.6%) patients.But there is no statically significant relationship between presence of PTS and Doppler findings. In addition to these, there is no statically significant association between PTS and presence of reflux-trombosis at any vessel in the affected leg, but there is a correlation between severe PTS and reflux(r=0.224, p:0.096).VCSS have positive correlation with the presence of reflux(p:0.041, r:0.224). VEINES-QoL/Sym,VCSS and BSAS were significantly worse in patients with PTS.(Table1)

Conclusion: In this study, we found that PTS in lower extremity develops in more than half of the patients with VBD during follow-up, and didn't found any predictor factor for develeopment of PTS. About one third of patient with PTS had severe PTS. PTS is an important clinical problem for physicians treating VBD in daily practice.It should be taken into account as much as preventing vascular relapses during follow-up of patients with VBD.

Table 1. Clinical and characteristic features of PTS

Disclosure of Interests: None declared
DOI: 10.1136/annrheumdis-2019-eular.4890

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A MONOGENIC DISEASE WITH WIDE RANGE OF SYMPTOMS: DEFICIENCY OF ADENOSINE DEAMINASE 2

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Background: Deficiency of adenosine deaminase 2 (DADA2) is an autosomal recessive autoinflammatory disorder caused by *ADA2* mutations.

Objectives: We aimed to investigate the characteristics of DADA2 patients along with the ADA2 enzyme levels.

Methods: 24 DADA2 patients who admitted to the Adult and Pediatric Rheumatology, Pediatric Haematology, and Pediatric Immunology Departments were included. All exons of the *ADA2* gene were screened by Sanger sequencing in all DADA2 patients. Serum ADA2 enzyme activity was measured by modified spectrophotometric method.

Results: 24 DADA2 patients were included; Group 1, 14 DADA2 patients with polyarteritis nodosa (PAN)-like phenotype; Group 2, 9 patients with Diamond-Blackfan anemia (DBA)-like features and one with immune deficiency. 14 PAN-like DADA2 patients did not have the typical thrombocytosis seen in classical PAN. Inflammatory attacks were evident in only Group 1 patients. Serum ADA2 was low in all DADA2 patients except one who was tested after hematopoietic stem cell transplantation. There was no significant difference in ADA2 levels between PAN-like and DBA-like DADA2 patients (Figure 1). ADA2 activities of heterozygote family members were about half the level of the control subjects. However, in heterozygote DADA2 patients, serum ADA2 levels were as low as the ones of homozygote DADA2 patients. *ADA2* mutations were affecting the dimerization domain in Group 1 patients and in the catalytic domain in Group 2 patients (Table 1).

Table 1. Molecular results of *ADA2* gene analyses in DADA2 patients (D, dimerization; C, catalytic)

Group number	Patient numbers	Mutation position	Mutation type	The affected domain of ADA2 protein
1	11	Exon 2: c.139G>A	Homozygous missense	D
1	1	Exon 2: c.139G>A/ Exon 2: c.140G>T	Compound heterozygous missense	D
1	2	Exon 2 c.139G>A	Heterozygous missense	D
2	1	Exon 4: c.620T>C/ Exon 9: c.1360G>C	Compound heterozygous missense	C
2	2	Exon 7: c.1072G>A	Homozygous missense	C
2	1	Exon 4: c.629delT/ Exon 2: c.144_145ins	Compound heterozygous Del/ins_frameshift and non-sense	C
2	1	Exon 10: c.1445A>G	Homozygous missense	C
2	1	Exon 4: c.680-681delAT	Homozygous deletion-Frameshift non-sense	C
2	1	Exon 9: c.1367A>G	Homozygous missense	C
2	2	Exon 6: c.916C>T	Homozygous non-sense	C
2	1	Exon 9: c.1392_1393insG	Homozygous missense	C

Conclusion: We suggest that enzyme activity of ADA2 should be assessed along with genetic analysis since there are heterozygote patients with absent enzyme activity. Our data confirms a possible genotype phenotype correlation where dimerization domain mutations are associated with a PAN-like phenotype whereas catalytic domain mutations are associated with hematological manifestations.

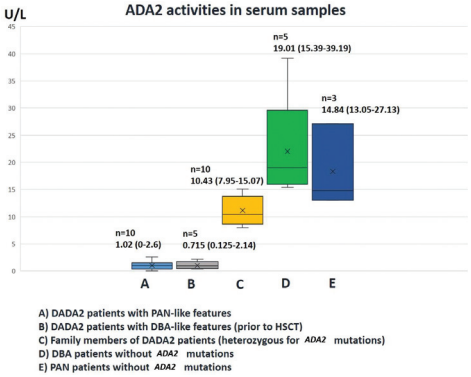

Figure 1

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Acknowledgement: This study was supported by Scientific and Technological Research Council of Turkey (TÜBITAK) with grant numbers of 315S192. Prof. Nurten Akarsu performed the molecular studies of DBA patients and special thanks to her for great support.

Disclosure of Interests: Ezgi Deniz Batu: None declared, Ekim Z. Taskiran: None declared, Hatice Asuman Özkara: None declared, Şule Ünal: None declared, Naz Guleray: None declared, Abdulsamet Erden: None declared, Omer Karadag: None declared, Fatma Gümrük: None declared, Mualla Çetin: None declared, Yelda Bilginer: None declared, Hafize Emine Sonmez: None declared, Deniz Cagdas Ayvaz: None declared, Ilhan Tezcan: None declared, Seza Özen Consultant for: Seza Ozen is receiving consultancy fees from Novartis, Speakers bureau: Roche

DOI: 10.1136/annrheumdis-2019-eular.584