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KLIPPEL-TRENAUNAY-WEBER SYNDROME: A CASE PRESENTATION

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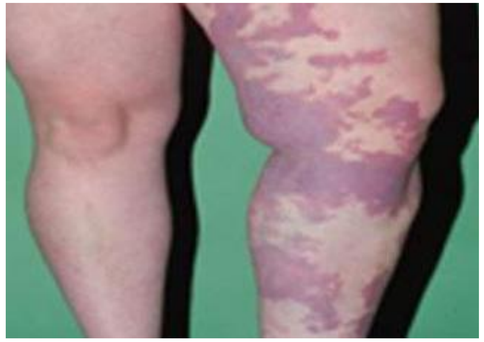
PURPOSE / OBJECTIVES

Klippel Trenaunay Webber Syndrome (KTWS) is a rare congenital disorder characterized by asymmetric limb hypertrophy, usually of the lower limbs, as well as vascular anomalies and capillary malformations. The aim of our study is to present the laboratory findings of a KTWS case.

MATERIALS & METHODS

A 20 year old female patient was presented at our laboratory for routine examination. We performed biochemical, haematological and haemostasis tests, capillary serum protein electrophoresis and immunofixation, as well as tests for the detection of autoantibodies.



	TEST	RESULT	UNITS
BLOOD COUNT	RBC	4,0	cells/μL
	HB	11,2	g/dL
	HCT	34,0	%
	PTL	112,0	cells/μL
HEMOSTASIS	PT	11,6	sec
	APTT	44,6	sec
	D-DIMER	21,9	mg/L
BIOCHEMISTRY	TBIL	2,07	mg/dL
	DBIL	0,43	mg/dL
	LDH	303,0	U/L
	CPK	472,0	U/L
	CRP	20,5	mg/L
	FERRITIN	231,0	ng/mL
SERUM PROTEIN ELECTROPHORESIS	ALBOUMIN	58,0	%
	α1-globulin	3,2	%
	α2-globulin	9,5	%
	β-globulin	13,7	%
	γ-globulin	15,6	%
AUTOANTIBODIES IFA	ANA	NEGATIVE	
	ANTI-DNA	NEGATIVE	
	ANTI-ENA	NEGATIVE	
	P-ANCA	NEGATIVE	
	C-ANCA	NEGATIVE	

RESULTS

Her complete metabolic panel and complete blood count were normal. Serum protein electrophoresis and immunofixation were normal too. CRP and ferritin levels were 92.2mg/L and 214.0 ng/mL respectively. Coagulation test results were PT 14.4 sec, aPTT 33.8 sec, INR 1.27, D-Dimer 31.87mg/L and Fibrinogen 96mg/dL.

SUMMARY/CONCLUSION

KTWS is rare, genetic disorder, associated with the translocation at t(8;14)(q22.3;q13). Its incidence and prevalence are not known. There is no apparent ethnic or gender predilection. Diagnosis of KTWS is mainly clinical. There are no specific laboratory tests. Tests should focus on evaluation of the type, extent and severity of the malformations. In this case report, CRP and Ferritin levels are elevated due to chronic inflammation. Moreover, D-Dimer levels were elevated due to deep vein thrombosis in the patient‘s legs.