

# Thrombelastography as screening test for the diagnosis of Scott Syndrome

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## Background

Scott syndrome (SS) is a rare bleeding disorder, characterized by impaired platelet procoagulant activity (PCA). The laboratory tests to diagnose this disease are available in specialized laboratories, and sometimes only for research purposes.

## Case report

A 35-year-old male patient was referred to our centre for diagnosis of hemostasis disorder. He had had a history of bruises and hematomas since childhood, one of them treated with surgical procedure. Blood transfusion was required after postectomy and crural hernioplasty. His family history for bleeding was negative.

## Results

Laboratory investigation showed normal measurement of all coagulation factors, including vWF antigen/activity, evaluation of fibrinolytic system (alpha2-antiplasmin, plasminogen and euglobulin lysis time) and platelet aggregometry with ADP, ADR, arachidonic acid, collagen and ristocetin. (Table 1)

Test	Result	Reference Range
PT	16,3s	78% (70-100%)
Fibrinogen	293mg/dL	(220-496mg/dL)
Euglobulin lysis time	200min	(80-310min)
APTT	R=1,37	(0,84-1,21)
FVII	69%	(50-150%)
FX	105%	(50-150%)
FV	80%	(50-150%)
Plasminogen	114%	(55-145%)
α2-antiplasmin	120%	(89-112%)
50:50 mixtures tests	FXII=151%, FXI= 109% (50-150%) FIX=132% FVIII= 181% (50-150%) FXIII:Ag=94% (75-155%)	
D-dimer	223ng/mL	(<255)
FDP	negative	
Immediate: APTT	R=1,15	
2h incubation: APTT	R=1,16	
TT	21,9s	(17,4s)
FvW:Ag	245%	(50-150%)
FvW:Rco	162%	(50-165%)
Platelet aggregometry	with ADP, ADR, AA, collagen and ristocetin: normal.	

Thromboelastography (TEG) was hypocoagulant (Fig. 1A: prolonged R and K, reduced angle, MA), which led to consider abnormality of PCA. Then a serie of TEGs was performed, mixing total blood (patient or control) with platelet poor plasma - PPP (Fig. 2A and 2B) or platelet rich plasma - PRP (patient or control) (Fig. 3A, 3B and 4).

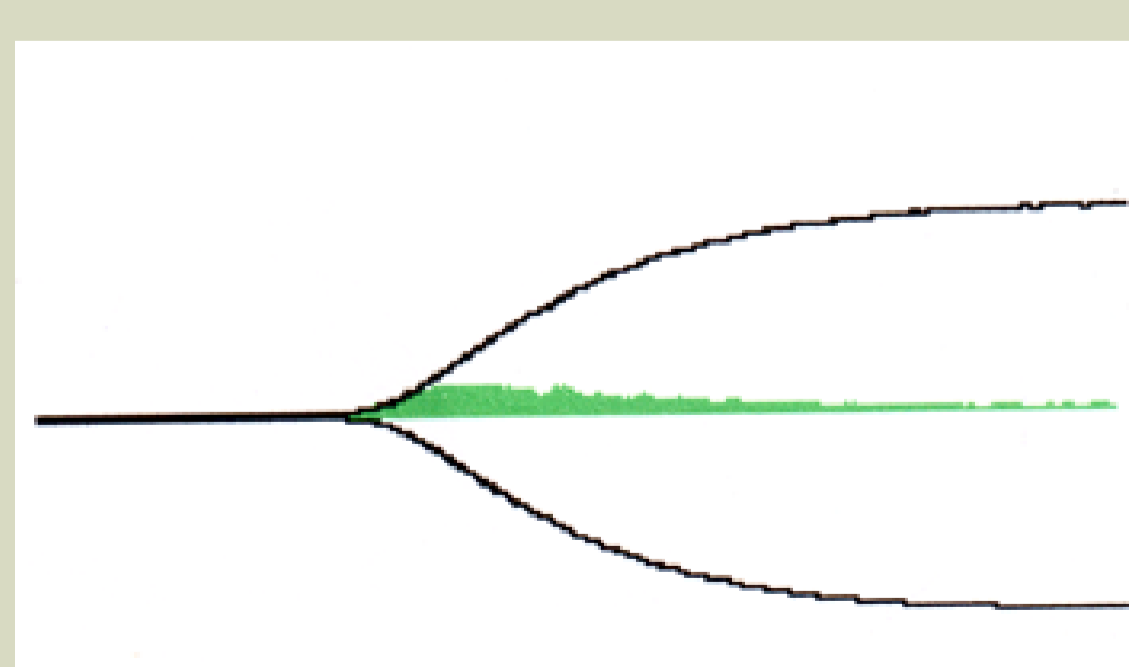


Fig.1 A: patient's TEG (total blood)  
R=18min (5-10), K=8,4min (1-3), Angle =24,3deg (45-74), MA=37mm (54-62)



Fig. 2 A: control TEG (total blood)  
R=7,8min, k=2,8min, Angle=53deg, MA=56,8mm

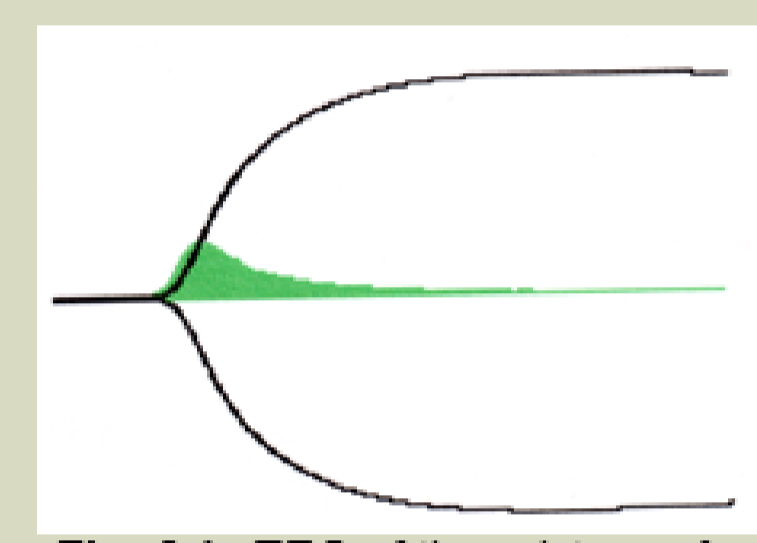


Fig. 2 A: TEG of the mixture of patient's total blood + PPP control  
R=9,1min, K=3,2min, Angle=50deg, MA=51,7mm

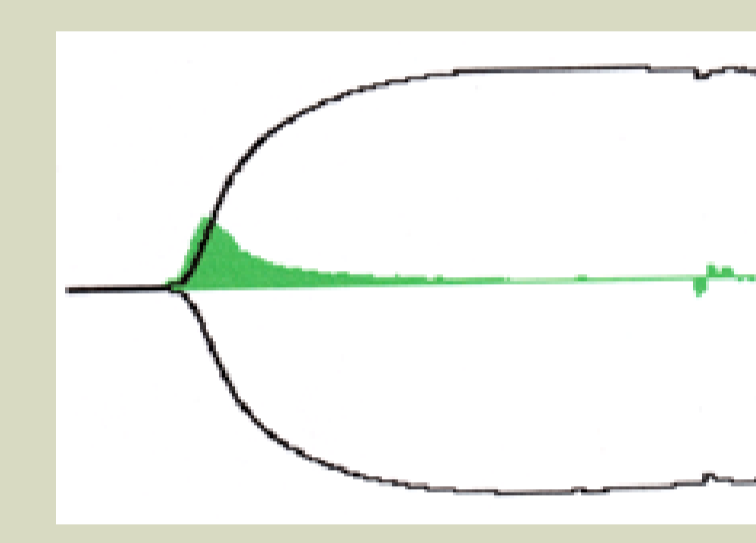


Fig. 2 B: TEG of the mixture of total blood control+ PPP control  
R=8,7min, k=2,5, Angle=54,9, MA=54,9

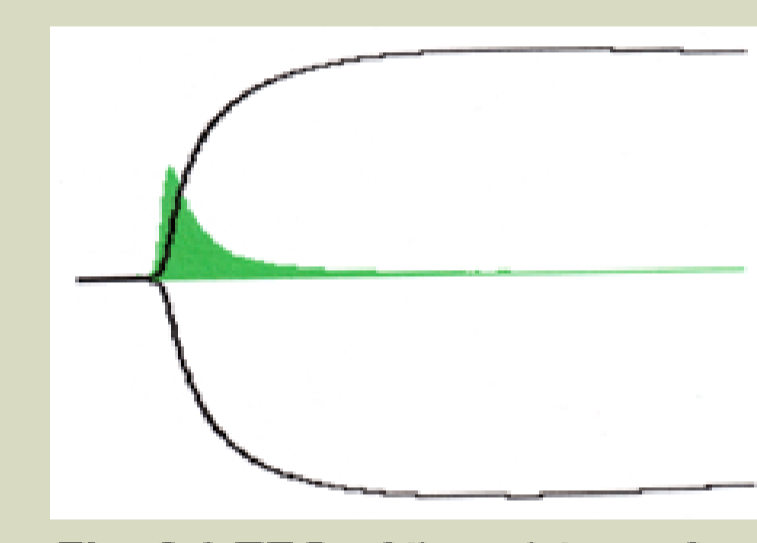


Fig. 3 A: TEG of the mixture of patient's total blood + PRP control  
R=6,2min, k=1,2min, Angle=64deg, MA=64,4mm

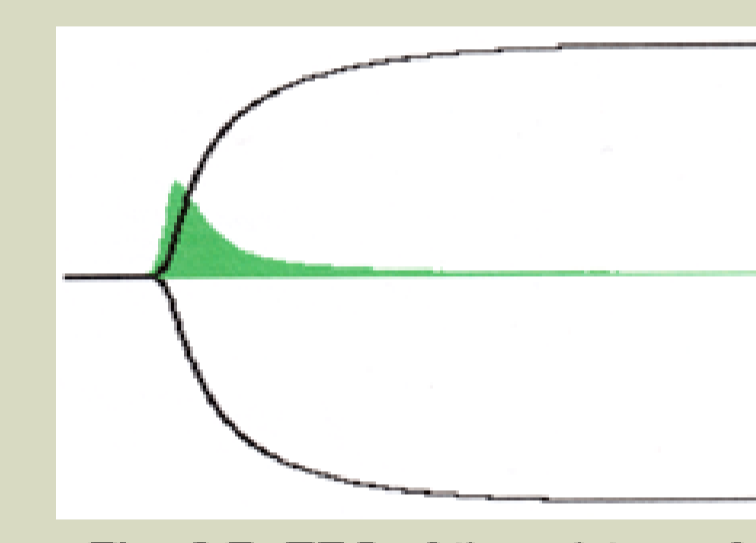


Fig. 2 B: TEG of the mixture of total blood control+ PRP control  
R=6,5min, k=1,5min, Angle=76,3deg, MA=67,9mm

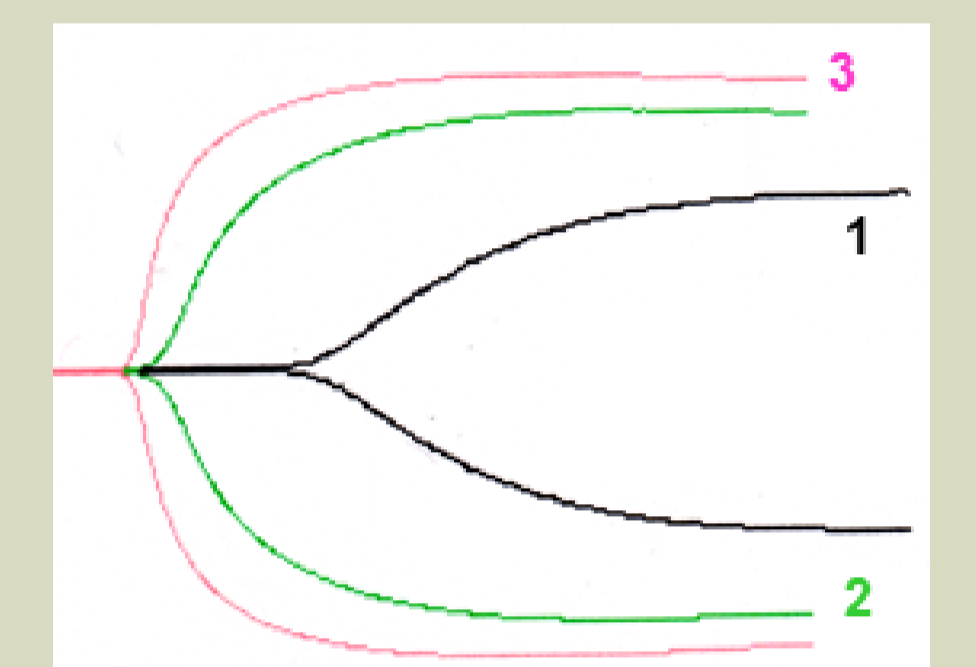


Fig. 4: TEG of 1- patient's total blood, 2- total blood control, 3- mixture of patient's total blood plus PRP control, showing correction.

	% POSITIVE FOR ANNEXIN V	
	CONTROL GROUP (N=4)	PATIENT
NO COLLAGEN	41±6	12
COLLAGEN 5µg/mL	56±4	18

	CONTROL GROUP (N=7) MEAN ± SD	PATIENT MEAN ± SD
LAG TIME (min)	7,6 ± 1,3	14,56 ± 2,86
ETP - Endogenous thrombin potencial - (nM)	1344,6 ± 130	727 ± 154,9
PEAK THROMBIN (Nm)	83,7 ± 8,6	36,78 ± 10,3
TIME TO REACH PEAK (min)	19,3 ± 3,4	25,33 ± 3,27

PLATELET RICH PLASMA= 250.000/mm<sup>3</sup>  
Reagents: Thrombinoscope - Netherlands - PRP-Reagent (1,0 pM of Tissue Factor)

## Conclusion

SS is a rare disease, and usually confirmatory tests are not part of the routine, even in specialized laboratories. TEG using mixtures of total blood and PRP of patient and control can be a simple and less expensive alternative method to screening impaired PCA in patients with bleeding disorders. In this case such approach helped in elucidating the diagnosis when sophisticated tests as annexin V and thrombin generation were not promptly available.

## References

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