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LABORATOIRE CMNN MONASTIR

Inherited combined deficiency of factor V and factor VIII: a case report managed by global tests and rotational thromboelastometry

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BACKGROUND

- ✓ Multiple coagulation factor deficiencies are a rare group of inherited hemostasis disorders with the simultaneous reduction of plasma activity of at least two coagulation factors.
- ✓ As consequence, the type and severity of symptoms and the management of bleeding/thrombotic episodes vary among patients.
- ✓ Combined deficiency of factor FV and FVIII and combined deficiency of vitamin K-dependent clotting factors comprise the vast majority of reported cases of familial multiple coagulation factors deficiencies.

CASE REPORT

- 38 years old male patient.
- **Medical history:** factor V constitutional deficiency discovered at the age of 8 years before an appendectomy with no personal history for spontaneous hemorrhagic disease.
- **Family history :** sister diagnosed with a combined deficiency of factor V and factor VIII discovered before delivery.
- **Reason for consultation :** factor V assay as part of a pre-operative assessment.

Laboratory findings

- **PT** = 68%
- **aPTT** = 53.9s/32s (ratio = 1,68), **kCTT** = 55.4s/29s (ratio = 1.91)
- **Fibrinogen** = 2.45 g/L
- **Factor V activity** =14.7%
- **Factor VIII activity** = 10.4%
- **Mixing study for both factors** ➔ factor deficiency
- **ROTEM ®** ➔ normal clot firmness (MCF) (**Figure 1**)

➔ **Inherited combined deficiency of factor V and factor VIII**

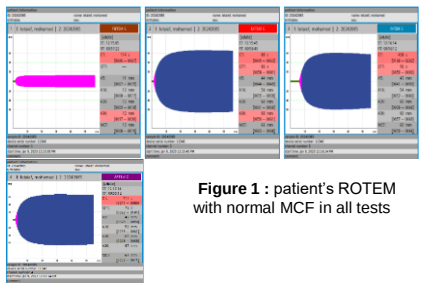

Figure 1 : patient's ROTEM with normal MCF in all tests

DISCUSSION

- ✓ Combined factor V and factor VIII deficiency (F5F8D) is a rare, autosomal recessive coagulation disorder.
- ✓ Cases are most concentrated in the Mediterranean, Middle Eastern, and South Asian countries, likely due to the prevalence of consanguineous marriages in these regions**.**
- ✓ The simultaneous decrease of plasma FV and FVIII, usually between 5% and 30%, is associated with a mild to moderate bleeding tendency **[1]**.
- ✓ The locus F5F8D maps to 18q21, between D18S849 and D18S1103 **[2].**
- ✓ Mutations in *LMAN1* or *MCFD2* genes lead to a defect in the intracellular transport complex LMAN1–MCFD2. As a result, the secretion pathway of FV and FVIII is disrupted.
- ✓ LMAN1 and MCFD2 form a Ca2+-dependent cargo receptor complex that functions in the transport of FV/FVIII from the endoplasmic reticulum to the Golgi apparatus.
- ✓ The recommended therapy includes fresh frozen plasma, which provides both of the deficient factors [3].

CONCLUSION

Although it's a rare occurrence, a factor V deficiency should prompt a search for a hidden factor VIII deficiency especially when PT and aPTT are both prolonged.

REFERENCES

[1] :,Zheng and al, combined deficiency of factor V and factor VIII, 2013
[2] : Arbez and al ; the locus of combined deficiency of factor V and factor VIII, 1997
[3] : Zheng and al, Combined deficiency of factor V and factor VIII is due to mutations in either LMAN1 or MCFD2, 2013