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LABORATOIRE CMNN MONASTIR

Rosenthal disease: a report of two cases

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BACKGROUND

- ✓ Factor XI deficiency (also called Rosenthal disease or hemophilia C) is a rare clotting disorder with prevalence of only 1:1000000; however, it is more prevalent in the Ashkenazi and Iraqi Jewish population.
- ✓ It is predominantly an autosomal-recessive trait, although some mutations follow an autosomal-dominant inheritance pattern.
- ✓ Patients with FIX deficiency demonstrate variable bleeding tendency despite severe deficiency (FXI activity <20%).

CASE REPORT

	Case (1)	Case (2)
Sex	69 years	29 years
Age	M	F
Medical history	Parental consanguinity and no personal or familial history for hemorrhagic disease	Ø
Reason of consultation	Exploration of an isolated prolonged aPTT fortuitously discovered in a pre-operative assessment	Exploration of an isolated prolonged aPTT fortuitously discovered before a non-scheduled Caesarean section following preeclampsia

	Case (1)	Case (2)
PT (%)	90	100
aPTT (s)	52.4/29 (ratio = 1.8)	75/29 (ratio = 2.58)
kCCT (s)	72.5/33 (ratio = 2.19)	99/33 (ratio = 3)
TT (%)	0.94	0.87
Fg (g/L)	2.45	-
Rosner index (%)	7	< 12
F VIII (%)	246	206
F IX (%)	108.6	132
F XI (%)	**1.4**	**0.2**
Mixing study	Correction	Correction

DISCUSSION

- ✓ The FXI gene is located on chromosome 4. Most cases of severe deficiency seem to follow an autosomal-recessive inheritance pattern; however, a dominant-negative effect has been observed in certain heterozygous genetic variants.
- ✓ Two genetic variants account for more than 90% of abnormal alleles : Glu117Stop (type II) and Phe283Leu (type III) **[1].**
- ✓ The bleeding phenotype does not correlate with the FXI activity level, with evidence of bleeding reported in heterozygotes with mild deficiency. This lack of correlation between bleeding risk and FXI activity levels poses a significant therapeutic challenge.
- ✓ Therapeutic challenges in managing patients with FXI deficiency include unpredictable bleeding that correlates poorly with FXI activity levels, lack of availability of FXI concentrate in many areas of the world, large volume of FFP required to achieve a hemostatic FXI activity level, and thrombotic risk associated with replacement therapy products **[2]** .
- ✓ Patients with XI deficiency should ideally be managed at a hemophilia treatment center. If this is not possible, they should be managed by a hematologist experienced in managing rare bleeding disorders. Multidisciplinary care is essential to ensure optimal patient outcomes **[3].**

CONCLUSION

Although factor XI deficiency is an underrecognized entity, it remains a rare bleeding disorder and it's often a diagnosis made fortuitously or prospectively after an excessive bleeding is encountered unexpectedly during surgical acts.

REFERENCES

[1] Lewandowska and al , Factor XI deficiency, 2021
[2] Wheeler and al, Why factor XI deficiency is a clinical concern , 2016
[3] Bolton-Maggs and al , Factor XI deficiency and its management, 2000