International Journal Dental and Medical Sciences Research
Volume 4, Issue 3, May-June 2022 pp 289-290 www.ijdmsrjournal.com ISSN: 2582-6018

A Rare Case of Factor 5 Leiden Mutation in Pediatrics Age Group

Dr Anna Roy, Dr Aditya Bhatt, Dr Praful Bambharoliya, Dr Yesha Patel
Resident doctor , Dept of Pediatrics GMC Surat
Resident doctor , Dept of Pediatrics GMC Surat
Assistant professor , Dept of Pediatrics GMC Surat
Resident doctor , Dept of Pediatrics GMC Surat

Submitted: 20-05-2022 Accepted: 30-05-2022

ABSTRACT
A 5yr old patient was referred to our hospital with c/o left sided knee swelling a/w pain and tenderness. Child was vitally stable, taking orally well with movement restriction due to pain. USG local part was s/o left knee joint effusion a/w left saphenous vein thrombosis amounting to absence of flow. Hence coagulation profile was sent. s/o Factor V Leiden mutation (heterozygous).

KEYWORDS : Factor V Leiden , thrombosis , LMWH , Venous Doppler

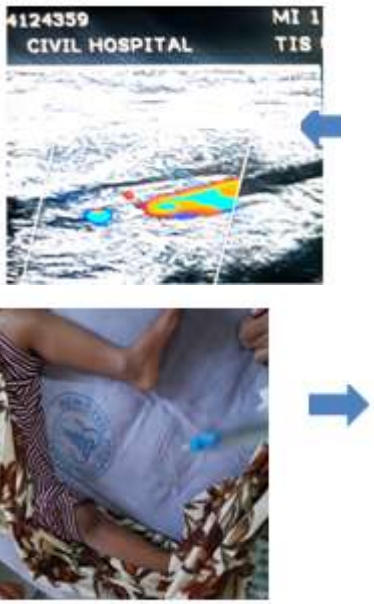
I. INTRODUCTION

Factor V Leiden mutation is a single nucleotide change at nucleotide 1765 within the Factor V gene which renders it resistant to inactivation by APC.[1]Heterozygosity for FVL mutation is the most common thrombophilia in individuals with venous thromboembolism [2] Heterozygosity of this gene increases lifetime risk for thrombosis by 7 times while homozygosity increase it by 20 fold. Diagnosed by functional coagulation test for APC resistance or by mutation analysis.

II. CASE DESCRIPTION

A 5 year old female child was referred to us with c/o left sided knee selling since 4 days followed by minor trauma at home. The child was vitally stable, on local examination left knee showed firm swelling with warmth tenderness associated with restriction of movement with no bony discontinuity. USG of bilateral knee joints done, and compared, revealed left sided knee joint effusion associated with venous thrombosis of great saphenous vein with absent flow. Complete coagulation profile was done along with other investigations, showed elevated CRP, D Dimer, LDH and ESR levels. Molecular analysis of factor V Leiden f5 R506q mutation by real time PCR showed pathological variant in heterozygous form. The child was started on LMWH subcutaneous injections along with other symptomatic managements. After 10 days of treatment

Doppler &sonography repeated s/o resolving thrombosis and partial flow through GSV. Hematoncologist opinion taken, enoxaparin tapered and stopped, warfarin started, parents counselled regarding risks of thromboembolism and regular follow up for future. Patient rendered asymptomatic with completely cleared thrombosis and reestablished blood flow through GSV by 3 weeks.



III. DISCUSSION

Factor V Leiden (FVL) is a point mutation of factor V resulting in an elimination of cleavage site in factor V and factor Va. This genetic defect leads to an increased risk of thrombosis especially in homozygous or pseudo-homozygous FVL mutations. Despite the increase in the risk of VTE, there is no clinical evidence that heterozygosity to FVL increases the overall mortality [3] The most common finding in individuals with FVL is a laboratory-only abnormality. Genetic testing is the

gold standard test and is indicated for those with a family history of FVL. Functional APC resistance assays cost less than genetic testing, but in rare cases, they can give a misleading falsely normal result which then requires confirmation via genetic testing. The first acute thrombosis is treated according to standard guidelines. Decisions regarding the optimal duration of anticoagulation are based on an individualized assessment of the risks for venous thromboembolism recurrence and anticoagulant-related bleeding.[4] In the absence of a history of thrombosis, long-term anticoagulation is not recommended for asymptomatic heterozygotic Factor V Leiden mutation patients.

IV. CONCLUSION

Due to unavailability of genetic testing at most health centres and financial constraints, clinician's expertise and judgement plays a pivotal role in the diagnosis of this condition. With well titrated anti coagulant therapy and routine follow up , patients with heterozygous Factor V Leiden mutation have excellent prognosis and life expectancy.

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