Utilization of obstetric ultrasound in the diagnosis of severe fetal ocular pathology associated with familial exudative vitreoretinopathy (FEVR)

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ABSTRACT

Introduction: FEVR is a rare inherited disorder leading to severe visual impairment and blindness. The primary pathophysiology of FEVR includes poor vascular differentiation and incomplete peripheral retinal vascularization. Subsequent ischemia and hypoxia can result in neovascularization and subsequently fibrovascular proliferation, falciform retinal folds, and retinal detachment. Clinical manifestations of FEVR are variable ranging from mild visual impairment to complete blindness at birth or during the first decade of life. Case Description: A 34-year-old, primigravida with severe visual impairment, strong family history of FEVR, and a normal 20 weeks anomaly scan, whose fetus was later found to have ocular pathology at 36 weeks. Ocular ultrasound showed evidence of retinal detachment with persistent hyaloid artery. A baby girl was later delivered at term and underwent an ocular laser ... and is currently diagnosed with bilateral FEVR. Discussion: Patients with strong family history of FEVR and a normal mid-trimester anomaly scan should be reassessed for fetal ocular changes in the third trimester in view, it is an evolving process. Prenatal diagnosis of FEVR via amniocentesis and molecular analysis is also essential to determine the inheritance of the genetic mutation. Prenatal diagnosis of FEVR via amniocentesis and molecular analysis though essential in determining the inheritance of the genetic mutation, an obstetric ocular ultrasound is fundamental to predict the severity of the condition in an affected pregnancy which allows prognostication of the unborn fetus and comprehensive counselling of the future parents.

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A proposed guide: Anti-Xa guided Low molecular weight heparin (LMWH dosing for pregnant mothers with mechanical heart valves as a safer option compared to the conventional weight based LMWH dosing

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ABSTRACT

Introduction: Achieving safe and effective anticoagulation among pregnant mothers with mechanical heart valves (MHV) remains challenging. Intravenous unfractionated heparin requires inpatient APTT monitoring while warfarin, especially doses beyond 5 mg is associated with a significant risk of fetal loss. Low molecular weight heparin (LMWH) has been proposed as an alternative anticoagulation although initial studies showed a significant risk of valve thrombosis among patients given weight based LMWH. Methods: We propose a precise strategy of Anti-Xa guided dosing of LMWH as a safer option in pregnancy, on par with some recent studies. This study retrospectively reviewed the trough and peak Anti-Xa levels of pregnant patients with MHV who were treated with weight-based dosing of LMWH in Hospital Tunku Azizah between September 2021 till April 2023 and analysed the doses required to achieve the desired Anti-Xa levels. Results: Eight patients were included in the study and we found that 1 mg/kg/BD dosing of LMWH was suboptimal among all of our patients (100%, n=8). The mean trough was 0.35 U/mL while the mean peak was 0.62 U/mL, far below the efficacy range. Our study showed that most patients required a mean LMWH dosing of 1.56 mg/kg/BD compared to conventional 1 mg/kg/BD dosing. Apart from being efficacious, it was also not associated with valve thrombosis, fetal loss, antepartum or postpartum haemorrhage. Conclusion: We propose a guide to suggest that treatment dosing of LMWH for pregnant mothers with MHV should be guided by peak and through Anti-Xa levels as a precise and safer strategy.