

P043

P043- Management of Gitelman syndrome in pregnancy: information resource for patients

Miss Rupa Kumar¹, Professor Fiona Karet Frankl¹

¹*University Of Cambridge, Cambridge, United Kingdom*

Introduction: Gitelman Syndrome (GS) is an inherited renal tubular disorder that presents mainly in adolescents and adults. It is caused by mutations in the gene encoding the thiazide-sensitive sodium-chloride cotransporter [1]. The thiazide diuretic-like phenotype includes hypokalemia, hypomagnesemia, metabolic alkalosis and hypocalciuria. Symptoms range from muscle cramps, paraesthesia and fatigue to seizures and life-threatening ventricular tachyarrhythmias [2]. Pregnancy poses a unique challenge to patients with GS due to physiological adaptations in kidney function, fetal demands and vomiting. This results in increased potassium and magnesium losses. Thus, GS symptoms tend to worsen during pregnancy, with a major impact on daily life. The risk of life-threatening complications is increased [3]. Currently, little information is available to these women regarding what to expect during pregnancy. Being a rare disease, clinicians involved in obstetric care of such patients may well be unaware of how best to manage them. We therefore sought to develop an information resource for patients detailing advice on managing the condition during pregnancy, also applicable to healthcare providers and eventually to be published on the rarerenal.org website [4].

Methods: First-hand experience was gathered by conducting interviews with four women with GS who had gone through pregnancy. The experiences of these women were used to inform the writing of patient information materials.

Results: The materials produced are written in the format of answers to questions that patients with GS are likely to have, related to all aspects of pregnancy. The answers explain the need for increased supplements for GS during pregnancy, the rationale for more frequent blood tests, and symptoms for which to seek urgent medical attention. We suggest that patients carry documentation explaining GS in the event of an emergency hospital admission. This resource emphasizes a collaborative approach to management involving a multi-disciplinary team.

Discussion/Conclusion: This patient resource aims to empower women with GS in pregnancy, by providing accessible information during what can be a difficult time in their lives. The resulting information resource will be used as a reference for healthcare providers.