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Women with Genetic Diseases and Obstetric Care

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COMMENTARY

Pregnant women with genetic illnesses are becoming a more relevant and essential topic. Advances in assisted reproductive technologies, as well as improvements in early detection and therapy of genetic illness, have influenced pregnancy rates in a group of women who might not have been able to conceive otherwise. Pregnant women with complicated health issues, such as genetic illnesses, require a multidisciplinary approach to their care. To enhance maternal and foetal health, important issues should be addressed before conception, during pregnancy, and during the postpartum period. Counseling about the risk of inheritance in offspring and prenatal diagnosis alternatives, if available, should also be reviewed.

Disorders of Metabolism

Inborn errors of metabolism are uncommon illnesses caused by the build-up or deficiency of a metabolite as a result of faulty enzyme function or the lack of essential enzymes. Early detection and treatment are critical for better outcomes. Due to the particular prenatal and obstetric difficulties connected with phenylketonuria and fatty oxidation disorders, this paper examines them.

Phenylketonuria (PKU) is an autosomal recessive illness characterised by Phenylalanine Hydroxylase (PAH) enzyme insufficiency, resulting in an inability to tolerate the necessary amino acid phenylalanine. Pregnancy among women with PKU is becoming more prevalent, thanks in part to earlier detection through newborn screening, which allows for improved treatment and outcomes. PAH gene mutations, and so therefore PKU, are most common in people of Northern European ancestry, where the condition affects 1 in 10,000 people. For phenylalanine metabolism, enough PAH enzyme activity is required. In affected individuals, eating phenylalanine causes a accumulation of phenylalanine and its metabolites. These metabolites are neurotoxic, and if not addressed, they can cause microcephaly, epilepsy, and intellectual impairment. Vomiting, irritability, tiredness, and increased tone are common disease symptoms in children. An increase in phenyl acetic acid, one of the phenylalanine metabolites, has been linked to a musty odour in people affected. If left untreated, phenylalanine levels can rise to 20 times normal, resulting in diminished skin and hair pigmentation due to tyrosinase suppression. Reduced myelin development and synthesis of various neurotransmitters result in neurological impairments. The severity of the condition is affected by treatment adherence as well as the genotype of the PAH.

In the United States, newborn screening has proven to be quite efficient in detecting PKU in infants within the first week of life and initiating therapy before irreparable neurologic impairment develops. The goal of treatment is to keep plasma phenylalanine levels between 120 and 360 mol/L (2-6 mg/dL) by following a low-protein, low-phenylalanine diet supplemented with a phenylalanine-free medical formula. PKU symptoms can be minimised or eliminated with proper adherence to a treatment programme initiated before the age of three months. Although diet changes are the mainstay of treatment, the FDA's approval of Sapropterin adds another treatment alternative. In patients with Hyperphenylalaninemia (HPA) due to BH4-responsive PKU, sapropterin lowers blood Phe levels. With continuous exposure to excessive phenylalanine levels, neurocognitive deterioration and behavioural problems can emerge even in later childhood and adulthood [1-6].

Women with PKU who are considering becoming pregnant should be advised on where it is of treatment adherence both before and during pregnancy. Plasma phenylalanine concentrations of 120-360 mol/L (2-6 mg/dL) for at least

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3 months before to conception are the treatment goals. Women should be monitored regularly by a nutritionist during pregnancy, as protein and dietary phenylalanine requirements fluctuate with gestational age. To achieve correct weight increase during pregnancy, diet should be maintained.

Women with genetic diseases are reaching reproductive age at an increasing rate, and many of them become pregnant. While some women can handle pregnancy and have positive outcomes, others may face a significant risk of morbidity and mortality as a result of their pregnancy. Management requires a diverse strategy that pays close attention to specific risks. Preconception consultation should ideally be indicated to examine maternal, perinatal, and neonatal hazards, as well as the risk of offspring recurrence.

REFERENCES

1. Chetty S, Norton ME (2017) Obstetric care in women with genetic disorders. Best Pract Res Clin Obstet Gynaecol 42: 86-99.
2. Kadir RA, Davies J, Winikoff R, Pollard D, Peyvandi F, et al. (2013) Pregnancy complications and obstetric care in women with inherited bleeding disorders. Haemophilia 19(4): 1-0.
3. Giammarile F, Bozkurt MF, Cibula D, Pahisa J, Oyen WJ, et al. (2014) The EANM clinical and technical guidelines for lymphoscintigraphy and sentinel node localization in gynaecological cancers. Eur J Nucl Med Mol Imaging 41(7): 1463-1477.
4. Motoshima S, Irie H, Nakazono T, Kamura T, Kudo S (2011) Diffusion-weighted MR imaging in gynecologic cancers. J Gynecol Oncol 22(4): 275-287.
5. Nazareth SB, Lazarin GA, Goldberg JD (2015) Changing trends in carrier screening for genetic disease in the United States. Prenat Diagn 35(10): 931-935.
6. Moos MK, Cefalo RC (1987) Preconceptional health promotion: a focus for obstetric care. Am J Perinatol 4(1): 63-67.