Maternal Phenylketonuria

Phenylketonuria is an autosomal recessive metabolic disorder caused by a mutation in the enzyme phenylalanine hydroxylase. This reaction requires tetrahydrobiopterin as a cofactor, and decreased tetrahydrobiopterin can also cause decreased activity of phenylalanine hydroxylase. For women with phenylketonuria, it is important for the health of their children to have low phenylalanine levels before and during pregnancy, because high levels of phenylalanine can cross the placenta and affect the developing fetus. Lack of proper diet during therapy, like eating high amounts of phenylalanine containing NutraSweet, can cause congenital heart disease, growth retardation, intellectual disability and microcephaly.



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Pathophysiology

Lack of Proper Diet During Pregnancy; Like NutraSweet
Mom eating Nutrasweet packet
NutraSweet is an additive high in phenylalanine, and a lack of a proper diet during pregnancy can cause symptoms in the developing fetus. For women with phenylketonuria, it is important for the health of their children to have low phenylalanine levels before and during pregnancy, because high levels of phenylalanine can cross the placenta and affect the developing fetus.

Signs and Symptoms

Congenital Heart Defects
Heart
Excess phenylalanine can disrupt cardiac development and cause congenital heart defects in the newborn.

Growth Retardation
C-clamp
Infants with maternal phenylketonuria fail to grow at an appropriate rate, and also fail to attain early developmental milestones.

Intellectual Disability (Mental Retardation)
Book Covered in Tar
Infants with maternal phenylketonuria can display severely impaired cognitive functioning with deficits in two or more adaptive behaviors.

Microcephaly
Small-head
A neurodevelopmental disorder in which the head circumference is more than two standard deviations smaller than the average circumference for the person's age and gender. It is a common clinical feature of infants with maternal phenylketonuria.

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