# GENE Seeker

You know the risk of your child being a carrier of a genetic disease.

# Phenylketonuria

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## What is Phenylketonuria?

Severe form of phenylketonuria (PKU) due to phenylalanine hydroxylase deficiency, an inborn error of amino acid metabolism, characterised in untreated patients by severe intellectual deficit and neuropsychiatric complications.

Late diagnosed patients present mostly with progressive developmental delay associated with severe signs including stunted growth, microcephaly, seizures, tremors, eczema, vomiting, musty odor, and subsequently behavioral (hyperactivity) and motor disorders. Untreated patients develop profound, permanent intellectual impairment and deterioration of cognitive performance and motor skills. Demyelination and decreased dopamine, norepinephrine, and serotonin production have been found in patients who do not pursue dietary restrictions into adulthood. Complications later on include exaggerated deep tendon reflexes, tremor, and paraplegia or hemiplegia. In treated patients, clinical signs vary based on treatment and diet compliance, and may include psychiatric disorders such as attention deficit-hyperactivity disorder and depression.

## What is the next step if I'm a carrier of Phenylketonuria?

If you are found to be a carrier of Phenylketonuria, it is important that your partner be tested for the same genetic disorder.

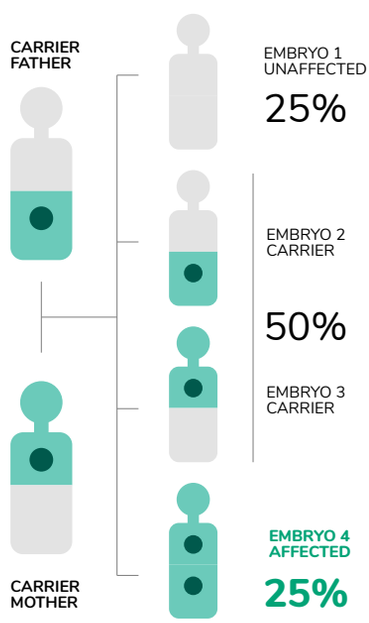
## What if my partner is not a carrier?

If your partner's test for Phenylketonuria, the chance to have an affected child is low. However there is currently no test able to detect all existing mutations, so there is always a residual risk that the person who has done the test is a carrier of other less frequent mutations.

## What if both me and my partner are carriers of Phenylketonuria?

When both parents are carriers ofPhenylketonuria, the probability of having a child with Phenylketonuria is 25%.

We recommend that you discuss your results with your doctor or genetic counselor in order to know more about reproductive options.




If both you and your partner are carriers, speak with your doctor or genetic counselor about reproductive options.

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