Hyperphenylalaninemia

- **increased levels of phenylalanine in the blood**
- AR hereditary, gene is on 12. chromosome, incidence is 1:10 000

Types

- *phenylalanine hydroxylase defect* - classical phenylketonuria (PKU)
- *dihydrobiopterin reductase defect* - atypical phenyketonuria
- *dihydrobiopterin biosynthesis defect* (phenylalanine hydroxylase cofactor) - atypical phenylketonuria
- phenylalanine increased only to 0.6 mmol / l is called "hyperphenylalaninemia"

Pathogenesis

The pathogenesis of HPA is the same as for other types, but it does not have nearly the same effect as **classical phenylketonuria** and is essentially disease-free.

Thus, hyperphenylalaninemia is only an increased amount of phenylalanine in the blood of a patient's normal diet.

Screening

🔎 *For more information see Neonatal screening.*

we use capillary blood from foot 2. − 3. day after birth

- at the time of collection, the newborn should have been on dairy food for at least three days
- when the child is released from the maternity hospital on the 2nd or 3rd day - we will take the screening, but after the release the practitioner will ensure the collection of capillary blood for a control examination
- from 1 October 2009, **newborn neonatal screening** is performed using the tandem mass spectrometry method
- formerly: Guthrie's method - we have Bacillus subtilis and its competitive inhibitor on agar, we add blood, in the presence of Pha it starts to grow

Blood testing of a two-week-old newborn for phenylketonuria

Diagnosis

- diagnosis can be made from the blood at the metabolic and molecular level, enzymatic examination from liver biopsy is not needed for diagnosis or treatment, but all children with PKU need to be examined **pterin metabolism** to differentiate forms

Links

related articles

- Phenylketonuria
- Neonatal screening
- Phenylalanine

External resources

- Fenylketonurie (česká wikipedie) (https://cs.wikipedia.org/wiki/Fenylketonurie%7C)
- Fenylalanin (česká wikipedie) (https://cs.wikipedia.org/wiki/Fenylalanin%7C)

Source

- BENEŠ, Jiří. Study materials [online]. © 2007. [feeling. 2010-04]. < http://www.jirben.wz.cz/ >.

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