INFANTILE HYPOLACTASIA: a case with challenging diagnosis

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Introduction

Lactose intolerance is the inability to metabolize lactose, because of the lack of enzyme lactase in the digestive system. There are known three major types of lactose intolerance. We report a patient with (adult type) of hypolactasia who was picked up by selective screening for galactosemia [1].

Methods

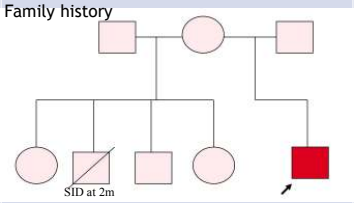
Urinary galactose/galactitol content was evaluated with Shimadzu HPLC system which was equipped with Aminex-HPX87H column and refractive index/ultraviolet-visible spectrophotometrical detectors. Native urine was analysed (no pretreatment, except filtration through 0.45 µm membrane filter)[2].

Case report

Case history

- Patient was born at term with birth weight 3360g and Apgar score 9/10;
- Meteoristic stomach pains since birth;
- By the age of 3 months the patient had been referred to our hospital 8 times due to irritability and meteorism of unknown origin.

Family history



At 2.5 months of age

- The urinary screening for metabolic disease was done.
- **Benedict's reaction was mildly positive (+).**
- We suggested urinary quantitative sugar analysis, which showed galactose excretion 334 mmol/mol cr (normal <377 – upper normal limit); no galactitol; lactose 44.3 mmol/mol cr (normally nd).

At 3.5 months of age

- He was admitted again with similar clinical problems
- Repeated sugar HPLC analysis:
  - **galactose in urine was increased 782 mmol/mol cr (normal <377);**
  - **galactitol 597 mmol/mol cr (normal <30);**
  - **lactose 20.3 mmol/mol cr (normally nd).**
- Lactate, ammonia and anion gap were normal.
- Urinary organic acid analysis showed also galactose conjugates, but also not in big concentrations (similar peaks, only much bigger, we see in treated galacosemia patients).
- Amino acids – normal.
- DNA analysis did not reveal Q188R mutation in GALT gene.
- He ate regular formula "Aptamil" on the moment of taking urine samples, which contained 6,2g/100 ml lactose.
- Lactose free diet was started → positive effect.

Patient at 3 months

Additional investigations

- Tandem MS analysis – normal.
- GALT activity 771 nmol/µmol Hb/hours (normal 597+/-51) (Maastricht).
- Galactose-1-phosphate <0.05 µmol/gram Hb (reference 0-0.05) (AMC = Amsterdam University Medical Centre).
- Uridine-diphosphate galactose epimerase activity 14.1 µmol/(hour.gram Hb) (normal 5.7-22.1) (AMC).
- Galactokinase (GALK) activity 44.8 nmol/min.gram Hb) (normal 28.5-78.1) (AMC).
- Sialotransferrines – normal (VUmc = Amsterdam Vrije Universiteit Medical Centre).
- Polyoles: pentosphosphate pathway normal, but galactose, galactitole and lactose excreation has risen (VUmc).
- We decided to test this child for hypolactasia as he responded positively to the lactose free diet.

He was found to be homozygous for c.-13910 C>T polymorphism C allele in MCM6 gene, which has influence to lactase florisine hydrolase (LCT) gene activity.

Diagnosis: (adult type) hypolactasia.

Discussion

In African population (adult type) of hypolactasia reveals clinically between 1 to 8 years and in Finnish population rarely before 5 years of age [3]. There is no data about the prevalence of hypolactasia in Estonia.

In case of hypolactasia the galactose content in urine is usually low. High content of galactose in urine is indication of normal activity of lactase in intestine [4]. Why in case in question the galactose content was high, we cannot explain. One possible reason may be the degradation of lactose in urine. But that does not explain the formation of galactitole that is a product of alternate pathway of galactose.

References

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Conclusion

Urinary galactose analysis might be a valuable tool for picking up infantile hypolactasia cases.

Acknowledgements

This work was supported by Estonian Science Foundation GARLA grant 8175.