Clinical Biochemistry 44 (2011) S64–S85

Contents lists available at ScienceDirect

Clinical Biochemistry

journal homepage: www.elsevier.com/locate/clinbiochem

12th ICB and 4th ICBMB (Abstracts of the 12th Iranian Congress of Biochemistry and 4th International Congress of Biochemistry and Molecular Biology, Mashhad, Iran, September 6-9, 2011)

Metabolic Inborn Diseases

Poster – [A-10-102-3]
Mutation screening of whole PAH gene in the PKU patients from North east of Iran

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Introduction: Phenylketonuria (PKU) is one of the most common inherited diseases causing mental retardation. It's inheritance pattern is autosomal recessive. The disease is characterized by deficiency of Phenylalanine hydroxylase (PAH) enzyme, which is due to mutations in the PAH gene. Molecular genetics diagnosis based on mutation identification in the affected families can leads to early treatment and prevention of mental retardation.

Material and Method: DNA was extracted from peripheral blood in 30 affected cases with classic form of PKU. The mutation was identified using PCR-RFLP and in some cases with direct sequencing of individual exons.

Results: While this is still an ongoing research, the results we had until now shows that the exon 11 represented 30% of mutations. The IVS10-11G>A is the common one. In this study one novel mutation was identified. The polymorphism L385L, c.1155G>C in the homozygout form was seen in 43% cases.

Conclusion: The mutations of the PAH gene in patients with classical phenylketonuria in Khorasan province were similar to that in other areas of Iran. Prenatal gene diagnosis for PKU by PAH gene sequencing might be efficient for most PKU families.

Keywords: PKU, Mutation, Mashhad

doi:10.1016/j.clinbiochem.2011.08.131

E Poster – [A-10-1025-1]
Prevalence of congenital hypothyroidism in newborns in Khorasan Razavi province, Iran

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Introduction: Congenital hypothyroidism important preventable cause of mental illness in newborns, but most babies are normal at birth. For these reasons screening tests for early detection of disease is used. Therefore, we wish to determine the prevalence of congenital hypothyroidism in Khorasan Razavi province during 2010.

Materials and Methods: In this cross-sectional study, from heel of the foot of almost all infants were born during 20 March 2009 and 20 March 2010 (n = 114,952) blood samples were taken during 3–5 days after delivery and transfer on paper Gatry and then samples transfer to Laboratory Health Center, Mashhad, Iran for measuring TSH. For subjects with THS higher than 5 mu/l, venous blood samples obtained using radioimmunoassay test for measuring TSH with T4 and T3 to confirm the diagnosis. The congenital hypothyroidism were diagnosed when in the neonatal period (1–4 weeks) these values were TSH ≥ 10mu / L and T4 < 6.5 μg/dl.

Results: In 109,964 infants the amount of TSH were < 5mu/l (95.66%), in 4664 newborn TSH levels were between 5–9.9mu/l (4.059%), TSH levels in 182 infants (0.177%) were between the 10–19.9mu/l and in 91 newborn (0.104%) TSH levels were more than 20mu/l. Among them, 168 (0.146%) neonates were confirmed their diagnosis as a congenital hypothyroidism.

Conclusion: Thereby, for every 684 infants one with congenital hypothyroidism was diagnosed. The prevalence of congenital hypothyroidism was one per 684 infants in Khorasan Razavi province.

Keywords: Congenital hypothyroidism, Neonatal screening, Thyroid stimulating hormone

doi:10.1016/j.clinbiochem.2011.08.132

E Poster – [A-10-1347-1]
Coronary artery angiographic changes in veterans intoxicated by mustard gas

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0009-9120/$ – see front matter.