Editorial

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21-Hydroxylase Deficiency: Newborn Screening in Iran?

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21-hydroxylase deficiency (21-OHD) accounts for the cause of 90-95% of congenital adrenal hyperplasia (CAH) cases. The world incidence of 21-OHD is 1:20,000 to 1:10,000 live births[1]. Prevalence of CAH trends to be high due to frequent consanguineous and first cousin marriages and underestimation because of stigmatization[2,3]. A range of clinical phenotypes including salt-wasting, simple virilizing and non-classic forms is emerged due to the variable residual 21-hydroxylase enzyme activity in CAH. Enzymatic defects in steroid biosynthesis pathway leads to accumulation of the metabolic precursors and shifting to androgen synthesis. Ambiguous genitalia appear in infant girls[4]. Basically, salt-wasting form occurs between first and third week after birth[4]. Because of nonspecific symptoms, an accurate diagnosis is often delayed so that males with classic form are at serious risk of morbidity (including neurological damage or intellectual disability) and mortality[4,5].

Newborn screening (NBS) for 21-OHD was performed for the first time in Alaska in 1977 and it is currently done in many European countries, USA, Canada and Japan[1,6,7]. The time-resolved, dissociation-enhanced, lanthanide fluorescence immunoassay (DELFIA) is used for 21-OHD NBS in most countries[8].

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Specificity and sensitivity of 21-OHD NBS are more than 99.5% and 92-100%, respectively[9]. Decreased morbidity and mortality associated with salt-wasting crises is the main objective of 21-OHD NBS [8]. Other important objectives are decreasing the time of sex assignment for infants with a virilized 46,XX karyotype, preventing precocious puberty and decreased final height in the simple virilizing form, and health improvement for the afflicted families[7]. 21-OHD NBS is usually performed before most babies with salt-wasting became symptomatic, so that it provides time for appropriate replacement therapy with hydrocortisone and fludrocortisone[8]. Molecular testing is currently performed in this country, but early detection would significantly decrease the costs, although no data is available worldwide on the cost-effectiveness of screening for this condition. Decreased hospitalization and decreased time to correct sex assignment have been documented in the screened populations. Liquid chromatography-tandem mass spectrometry, however, is more reliable and less costly than molecular testing[8].

In conclusion, CAH has all criteria for NBS[10]: 1) if undetected, it leads to high morbidity and mortality; 2) if detected early, an effective cheap treatment exists for the patients; 3) the 21-OHD NBS test would be efficient and reliable; and 4) the incidence of CAH is also high in our country[11]. However, a pilot study including approximately 10,000 infants is recommended to decipher incidence of disease and cost-effectiveness of the test; so that health professionals decide whether to perform 21-OHD NBS or not.

Key words: 21-hydroxylase Deficiency; Congenital Adrenal Hyperplasia; Screening

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