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Yenidoğan Tarama Programı

Newborn Screening Programs

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Abstract

Newborn screening programs are carried out in the world and in our country in order to diagnose the symptoms of treatable or controllable metabolic, endocrine and genetic diseases before their symptoms appear, to prevent long-term serious problems by treating them and to reduce treatment costs. In the development of these screening programs, diseases are included in the screening program, taking into account the criteria determined such as disease, test and treatment for their validity, effectiveness and suitability. By taking a blood sample for Guitre test from every newborn at postnatal 36-72 hours, it is aimed to reduce mortality and morbidity rates with early diagnosis and treatment of diseases such as phenylketonuria, congenital hypothyroidism, biotidinase deficiency, cystic fibrous and congenital adrenal hyperplasia. In addition, hearing, vision, congenital heart diseases and developmental pelvic dysplasia are included in newborn screening programs.

Keywords: Newborn, screening program, screening tests

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Neonatal screening programs include the tests performed to determine the diseases in the newborn period (İçke & Ekti Genç, 2017:186). When the disease is included in the screening program, criteria such as the frequency of occurrence, the severity of the problems that may arise when untreated, the extent to which these problems can be prevented when treated, and the cost of treatment and screening are evaluated (Beşirik & Canbulat, 2019:323; Caggana et al., 2013:14). Phenylketonuria (PKU), congenital hypothyroidism, biotidinase deficiency, cystic fibrosis and congenital adrenal hyperplasia are screened in our country with the newborn screening program. Apart from these, hearing, vision, congenital heart diseases and developmental hip dysplasia were included in the screening program (Güner ve Güner, 2017:204).

Phenylketonuria: It is an inherited metabolic disease due to the deficiency of the enzyme phenylalanine hydroxylase. Turkey is one of the countries most frequent incidence of PKU is a newborn 3000-4000. Consanguineous marriages increase the incidence of this disease, which shows autosomal recessive inheritance (İçke and Ekti Genç, 2017:186; Beşirik & Canbulat, 2019:323). Babies with phenylketonuria are normal at birth. In babies who are not diagnosed in the early period or who are fed a diet containing phenylalanine, symptoms occur after the first months. Due to the reduction of pigmentation, most children have fair skin, fine blond hair and blue eyes. Growth and growth retardation, vomiting, irritability, hyperactivity, convulsions, atopic dermatitis and a musty odor in urine are other symptoms. Mental retardation is seen in untreated children (Albrecht, 2009:414; İçke and Ekti Genç, 2017:186; Beşirik & Canbulat, 2019:323; Strisciuglio & Concolino, 2014:1007). Normal level of phenylalanine is 1.6-2 mg / dl. If the phenylalanine level is above 4 mg / dl, the diagnosis of phenylketonuria is made (van Wegberg et al., 2017:1). In the treatment, the normal development and social integration of children should be ensured by restricting phenylalanine (Strisciuglio & Concolino, 2014:1007).

Congenital Hypothyroidism: Congenital hypothyroidism is caused by developmental disorders of the thyroid gland or congenital thyroid hormone deficiency. Incidence increases in Down syndrome, which is one in 3,500-4000 live births. Early diagnosis and treatment is important in untreated children due to growth and development retardation and mental retardation (Beşirik & Canbulat, 2019:323). In babies with congenital hypothyroidism, hoarse crying, openness in the posterior fontanel, drop in body temperature, dry skin, a large tongue, feeding difficulty, abdominal distension, constipation, umbilical hernia, prolongation of neonatal jaundice are observed. In addition to these, while delay and structural disorders are observed in the emergence of teeth, their necks are short and thick, facial expressions are dull,

arms and legs are short and hypotonic (Rastogi & LaFranchi, 2010:1). If the TSH (Thyroid Stimulating Hormone) cut-off value is above 20 mU / ml in the Guthrie test, newborns are recalled. T4 level is measured with 20-40 mU / ml. The aim of the treatment is to start thyroid hormone treatment as soon as possible and to return the thyroid hormone level to normal (Rose and Brown, 2020:2229).

Biotinidase Deficiency: It is an autosomal recessive disorder caused by deficient biotinidase enzyme activity. Generally, it gives clinical symptoms in the first 3-6 months. Biotinidase deficiency occurs in one in 60000 live births. Symptoms and signs seen in partial and severe biotinidase deficiency vary. Partial biotinidase deficiency is a milder form. In severe biotinidase deficiency; Seizures, weak muscle tone (hypotonia), respiratory problems, hearing and vision loss, movement and balance problems (ataxia), skin rashes, hair loss (alopecia) are seen. In addition, developmental delay can be seen in affected children (Wolf, 2016:1). The diagnosis of this disease can be easily made with newborn screenings and early findings can be prevented. It is treated with oral biotin.

Cystic Fibrous: It is an autosomal recessive genetic disease characterized by dysfunction of the external glands. Physical changes occur in the respiratory, gastrointestinal, skeletal and reproductive systems due to the external glands being affected. Since abnormalities occur in the transport of chlorine and water in the epithelial cells, the secretions of the sweat glands, gastrointestinal system, respiratory system and other external glands darken and blockages occur in the ducts (Yıldırım Sarı & Bektaş, 2018:99). The frequency of CF in newborns according to societies varies between 1/2500 and 1/20000 (Üstü & Uğurlu, 2016:239). Meconium ileus occurring in the first 48 hours in the newborn should suggest cystic fibrosis. Abundant stool with oily, bad smelling is observed. Respiratory distress, cough and hypoxia are observed due to the thick and sticky secretion causing obstruction in the airways. Accordingly, respiratory tract infection develops. Excessive excretion of chlorine with sweat glands leads to fluid and electrolyte imbalance. For diagnosis, sodium and chloride levels should be determined by sweat test (Yıldırım Sarı & Bektaş, 2018:99; Davis, 2006:475). In diagnosis, the chlorine level in the sweat test is evaluated. Treatment includes maintenance of airway patency, infection control, providing and maintaining the highest level of nutrition, breathing exercises and prevention of intestinal obstruction (Yıldırım Sarı & Bektaş, 2018:99).

Congenital Adrenal Hyperplasia (CAH): It is caused by a deficiency of one or more enzymes necessary for the synthesis of cortisol and sometimes aldesteron. The most common type of CAH is the classical type and its incidence is one in 16,000. It shows autosomal recessive

inheritance (Merke & Bornstein, 2005:2125). CAH should be suspected in girls with abnormalities in the external genital organs such as enlargement of the clitoris, the appearance of the labia similar to the scrotum, and the absence of testicles. Genital organs are normal in male sex. In early childhood, the penis scrotum grows while the testicles remain small. Especially high levels of 17-OH progesterone are important in diagnosis (Speiser et al., 2010:4133). The essence of the treatment is to replace the missing hormones. In addition, surgical intervention may be required for abnormalities in the external genitalia.

References

Albrecht, J. 2009. Neuropsychological speedtest and blood phenylalanine levels in patients with phenylketonuria: A meta-analysis. *Neurosci Biobehav Rev,* 33, 414.

Beşirik, S., & Canbulat, N.Ş. 2019. Sağlıklı yenidoğan döneminde bakım. Diler Aydın, Yılda Arzu Aba (Eds), *Anne ve çocuk sağlığı* içinde (s. 323-343). İstanbul: İstanbul Tıp Kitabevleri.

Caggana, M., Jones, E.A., Shahied, S.I. Tanksley, S., Hermerath, C.A., & Lubin, I.M. 2013. Newborn screening: from guthrie to whole genome sequencing. *Public Health Reports*, 2(128), 14-19.

Davis, P.B. 2006. Cystic Fibrosis since 1938, *American Journal of Respiratory and Critical Care Medicine,* 173, 475-482.

Güner, Ş.K., & Güner, S. 2017. Gelişimsel kalça displazili çocuklarda tedavi yöntemleri. *Van Tıp Dergisi,* 24(3), 204-209.

İçke, S., & Ekti Genç, R. 2017. Topuk kanı örneği ile yapılan ulusal yenidoğan tarama testleri ve önemi. *The Journal of Pediatric Research*, 4(4), 186-90.

Merke, D.P., & Bornstein, S.R. 2005. Congenital adrenal hyperplasia, *The Lancet*, 365, 2125-2136.

Rastogi, M.V., & LaFranchi, S.H. 2010. Congenital hypothyroidism. *Orphanet Journal of Rare Diseases,* 5(17), 1-22.

Rose, S.R., & Brown, R.S. 2020. Update of newborn screening and therapy for congenital hypothyroidism. *American Academy of Pediatrics,* 2290-2303.

Speiser, P.W., Azziz, R., Baskin, L.S., Ghizzoni, L., Hensle, T.W., & Merke, DP. 2010. Congenital adrenal hyperplasia due to steroid 21-hydroxylase deficiency: An Endocrine Society Clinical Practice Guideline. *Journal of Clinical Endocrinology Metabolism*, 95(9), 4133–4160.

Strisciuglio, P., & Concolino, D. 2014. New strategies for the treatment of phenylketonuria (PKU). *Metabolites.* 4, 1007-1017.

Üstü, Y., Uğ & urlu, M. 2016. Ulusal erken tanı ve tarama programı: Kistik Fibrozis. *Ankara Medical Journal,* 16(2), 239-241.

van Wegberg, A., MacDonald, A., Ahring, K., Bélanger-Quintana, A., Blau, N., Bosch, A. M., … & van Spronsen, F. J. 2017. The complete European guidelines on phenylketonuria: diagnosis and treatment. *Orphanet Journal of Rare Diseases,* 12(1), 1-56.

Wolf, B. 2016. Biotinidase deficiency synonym: Late-onset multiple carboxylase deficiency. *GeneReviews*, 1-18.

Yıldırım Sarı, H., & Bektaş, M. 2018. Solunum yolu hastalıkları ve hemşirelik bakımı. Hicran Çavuşoğlu (Ed.), *Pediatri hemşireliği akıl notları* içinde; 99-113, Ankara:Güneş Tıp Kitapevi.