

Current situation of newborn screening for congenital hypothyroidism in China

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Neonatal screening for congenital hypothyroidism is an effective way to improve birth population quality which is carried out at the international level for nearly 50 years and carried out in China for 30 years, it is one of the most successful public health measure. Domestic and international data show that the development and promotion of neonatal screening for congenital hypothyroidism disease has social benefit to prevent and improve the health of children with mental retardation, to improve the quality of the population born. Neonatal screening is gradually popular today, with improvement in detection technology, systems technology to enhance and improve diagnosis and treatment, Chinese new born screening for congenital hypothyroidism thyroid level will rapidly increase. For 50 years, newborn screening has saved many infant lives, or eliminating the occurrence of severe mental retardation and their physical development disorder, brought joy to millions of households. Status of Chinese newborn screening as follows.

First, a brief history of neonatal screening for congenital hypothyroidism

China started to carry out newborn screening for congenital hypothyroidism later. In 1981, Shanghai and Beijing carried out the first screening for congenital hypothyroidism disease. after Shanghai, Beijing exploring sexual conduct after neonatal screening, other cities have been carrying out this work. Neonatal screening for congenital hypothyroidism gradually get the attention of government, national policy on neonatal screening were introduced: in October 1994, "People's Republic of China Maternal and Child Health Law" enacted, the provisions of "gradual development of neonatal diseases screening", first in the form of regulations to determine the status of neonatal screening in disease prevention; in October 2001, China began the implementation of "People's Republic of China Maternal and Child Health Law Implementation Measures", neonatal screening included in the technical services of maternal and child health; in 2004, the Ministry of Health issued a "neonatal screening technical specifications"; from June 1, 2009 implementation of "neonatal screening management approach", neonatal disease Screening management continue to be standardized. Currently, in most Chinese provinces to carry out newborn disease screening programs carried congenital hypothyroidism screening.

Second, congenital hypothyroidism disease screening purposes and principles selected

The purpose of neonatal screening for congenital hypothyroidism disease is to find serious, treatable disease in newborn children and

apply appropriate interventions, and when to avoid or ameliorate adverse outcomes. Currently, tandem mass spectrometry can detect a variety of diseases, some patients may have substantially detected incurable disease. It was in favor of an incurable disease screening because families of children can benefit from a clear diagnosis, but not likely to last not know the cause of death.

Neonatal screening conditions are met briefly summarized as the following four points: (1) Early diagnosis can allow infants to benefit (even some incurable disease, in favor of the family as a whole can benefit infants and young children). (2) Benefits may be a reasonable balance between the economic and other costs. (3) There is a reliable detection method for neonatal screening. (4) There is a satisfactory operating system to handle diagnostic tests, consultation, treatment and follow-up of patients detected.

Third, the prospect of congenital hypothyroidism disease screening

Neonatal screening has become one of the most successful public health policy since the start. This is largely due to advances in analytical techniques biomarkers of disease areas, the importance of the active participation of parents and community organizations in promoting people to be aware of the importance of disease and treat disease screening method. Challenges neonatal screening is required to ensure that the medical profession does not exceed the range of detectable cognitive disorders or have adequate resources and facilities to manage these abnormalities of the disease [1-23].

In recent years, the development trend of international neonatal screening and gradually increased to the center with tandem mass spectrometry screening, a comprehensive cost-effectiveness analysis showed that tandem mass spectrometry is to reduce the cost of neonatal screening [24]. In addition to improving the efficiency of tandem mass spectrometry detection, the false-positive rate was significantly lower, not only the newborn disease screening techniques to a new level, has become the development direction of newborn screening for inherited metabolic diseases.

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DNA microarray technology is currently used in neonatal screening so that the underlying disease due to mutations caused obtain early diagnosis, laid the foundation for further gene therapy. After the detection of biochemical markers have been mature during the traditional detect genetic diseases, gene level may be used as a further diagnosis, gene diagnosis such as congenital adrenal hyperplasia and cystic fibrosis [25]. To clear the gene mutations cause hereditary diseases, DNA testing is a priority level of inspection.

Application information integration system will rapidly integrate newborn screening laboratory data to the medical diagnosis and treatment agencies and public health authorities, effectively improve management efficiency at home and abroad which has become the focus of research [26]. Health authorities gradually strengthened the supervision of newborn screening systems, improve information, assess the service system of cooperation, cost-effectiveness analysis, knowledge of education and other aspects of the work [27]. Medical ethics of genetic testing and newborn screening expansion diseases involved cannot be ignored [28]. Screened children from childhood to adulthood, the system of medical care have gradually been concerned about.

Fourth, congenital hypothyroidism disease screening technology development

In recent years, a variety of analytical techniques have become routine laboratory neonatal screening technology used. Typically, screening strategies include determining an initial screening analytes and subsequent secondary sample testing analysis to improve its sensitivity and specificity. The initial screening technologies tandem mass spectrometry, colorimetric analysis test, fluorescence analysis test, enzyme or liquid chromatography and various immunological assays and other analysis, many of which can also be used for secondary analysis. With the improvement of existing technologies as well as new detection method of the present invention, it may be the number of disease detection screening will continue to increase.

Fifth, the cause of congenital hypothyroidism (CH)

Congenital hypothyroidism (congenital hypothyroidism, CH) is the most common endocrine disease of newborns, congenital hypothyroidism is mainly due to thyroid aplasia or hypoplasia, may be present in the body to inhibit the growth of thyroid cells immune globulin; followed by thyroid hormone synthesis pathway enzyme defect (an autosomal recessive genetic disease); congenital hypothyroidism incidence in all ethnic groups of approximately 1/3000. China CH prevalence of approximately 49.2/100,000 [29,30]. Sick newborns usually appear normal, it is difficult to find in the case of the absence of screening, but the delay in treatment will lead to mental disorders, developmental delay.

The congenital hypothyroidism screening methods; CH: each screening laboratory according to their own conditions, were taken to radioimmunoassay (RIA), enzyme-linked immunosorbent assay (ELISA), enzyme immunoassay fluorescence analysis (EFIA) and time-resolved immunofluorescence assay (DELFA) and other methods Determination of filter paper blood spots of thyroid stimulating hormone (TSH) concentrations. After 1998, CH screening mainly high sensitivity DELFA method, a few areas by ELISA and EFIA hair, RIA method has been basically no longer used. TSH positive cut value using 8-20 mIU/L range depending on the circumstances around the laboratory and kit, shear value than those who recalled review.

Sixth, China congenital hypothyroidism situation

China CH prevalence overall showed a rising trend, the prevalence was 24.5 in 1991 before/100,000, reached its peak in 2003 62.1/100,000, by 2006 dropped to 57.8/100,000. It is noteworthy that before 1998 CH prevalence has stabilized at a lower level, with an average of 19.7/100,000 from 1998 to 2006, CH prevalence rate increased an average of 51.8/100 000, mainly with improvement and recall of children suspected an increased detection methods. China's western region CH prevalence in the east and the Middle East, Central lowest prevalence.

Seven, congenital hypothyroidism treatment and follow-up case

CH is mainly due to thyroid congenital dysplasia, or many other factors by the presence of congenital defects which is caused by iodine deficiency and thyroid hormone synthesis. But for whatever reason, it must be made in thyroxine replacement therapy. Regular treatment is generally 2-3 years, the periodic review of thyroid function, thyroid B ultrasound or thyroid isotope scan, such as the withdrawal of temporary CH regular followup, such as permanent CH is to be lifelong replacement therapy, drug therapy is currently the main alternative L-thyroxine and thyroid is sodium, which is already widely used. Children with regular physical development, mental development and other assessments, and any time to adjust treatment.

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