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# Regional difference of genetic factors for congenitals cataract. The results of congenital cataract screening under normal pupil conditions for infants in Tianjin city

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**Abstract. – PURPOSE: To inquire into the morbidity of congenital cataract which affects the visual development of infants in Tianjin, find the proportion of cases caused by genetic factors, and to provide the baseline information for discovering amblyopia in an early stage so as to reduce vision disability.**

**MATERIALS AND METHODS: Babies no more than two months old were screened for congenital cataract under normal pupil conditions. The check-up was performed using ophthalmoscope and a portable slit lamp.**

**RESULTS: Among the 268,989 babies (537,978 eyes) being checked, 44 cases (75 eyes) of congenital cataract (13 unilateral cases and 31 bilateral cases) were detected and a morbidity rate of 0.016‰ was reported. 13 of the 44 babies (0.048‰) face difficulties in vision development. In the 44 positive cases, 7 had a family history of congenital cataract (15.91%), 8 (18.18%) had high risk factors and the other 29 cases (65.91%) reported no known reasons.**

**CONCLUSIONS: Babies eyes screening under normal pupil conditions is an effective, rapid and acceptable method for discovering congenital cataract cases. Unlike findings in other related literatures, we found that genetic factors accounted for 15.91% of the incidence of congenital cataract, while 65.91% cases can't be explained.**

*Key Words:*
Congenital cataract, Regional difference of genetic factors, Babies, Screening under normal pupil conditions.

## Introduction

Congenital cataract is the opacity of part or the entire lens of infant at birth or within first year of birth. It is an important cause of child blindness and amblyopia especially when taking place at the center of the visual axle. It not only affects vision but also influences visual functions of stare reflex, spatial vision, and color vision in children[1,2]. It is reported that approximately 10%-38% of childhood blindness is associated with congenital cataract[3]. Since congenital cataract is curable, early diagnosis and timely operation are crucial to reduce surgical complication, and to improve success rate. With funds from the Tianjin Municipal government, Tianjin Women and Children Health Care Center conducted a free check-up program in October 2008 to screen new-born babies in Tianjin for congenital cataract. To the best our knowledge it is the first time such a large scale screen conducted in China. We now present the method and results in this report.

## Materials and Methods

### Candidates Screen

Infants within 2 months who were born in Tianjin between October 15th, 2008 through October 15th, 2011 were screnned. Doctors participating in the program were experienced eye clinicians from local Women and Children Healthcare Organizations in Tianjin. They were well trained and obtained qualifications for the check-up program from the Tianjin Eye Hospital and the Ophthalmology Center of Tianjin Medical University. They continued regular repeated training and being annually examined for the screening procedure at the Tianjin Women and Children Health-care Center.

### Screening Location and Procedures

The total number of 18 screening institutions included Tianjin Women and Children Health-



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care Center and District and County-level Women and Children Health-care centers in Tianjin. It covers all the newborn babies born in Tianjin in the period.

When newborn babies aged 1-2 months were brought to Community Hospitals (the total number of 280) for vaccination, their parents were informed of the free-screening program and filled in the consent on a voluntary basis. The babies were then brought to the 18 Screening Institutions for a primary screen. At 6 months and 1 year old, all babies were rescreened at the Ophthalmology Department of the Tianjin Women and Children Health-care Center. Positive cases would be transferred to the specialized Department for Congenital Cataract in the Tianjin Eye Hospital for confirmation and treated accordingly. The detailed flow chart is shown as Figure 1.

According to the characteristics of the incidence of congenital cataract, we performed a primary screening and a rescreening.

### Primary Screening Methods

Infants within 2 months were placed on bed in the examination room, awake and in the supine position. Doctors first observed the lids and the conjunctiva with penlight in turn in the natural light. Secondly, an optokinetic nystagmus instrument was used to detect possible incidence of nystagmus and the direct and indirect light reflex of pupils also with a penlight. This is to test the function of the afferent and efferent neurological pathway responsible for pupillary reactions. Thirdly, reduce the brightness of the room into a dimmed room and then looking at the infants' pupil through a direct ophthalmoscope from 30 cm away using spot beam with a +2 diopter lens to observe the red light reflex of pupils. Finally, doctors hold portable slit lamps to examine the cornea and lens of the baby. A validated questionnaire survey of the parents was done to document a possible existence of congenital cataract risk factors[4,5], family history and mydriasis. Positive cases were simultaneously checked for leukocoria in differential diagnosis.

### Rescreening Methods

All infants in the study took part in a second and third check-up at 6 months and 1 year of age in the Ophthalmology Department of the Tianjin Women and Children Health-care Center. Mydriasis examination was done depending on results from previous pupil examinations.

### Gold standard for Diagnosing Congenital Cataract

Cloudiness of part or all of the lens of the baby at birth or in the first year after birth was considered the gold standard[4,5] for congenital cataract diagnosis.

### Data Management and Quality Control

For strict management and timely monitor, doctors of the 18 participating institutes inputted and uploaded outcome data and related information monthly into the Tianjin Maternal and Child Health Information Network. It was also where data reported in this study was from. The Tianjin Women and Children Health-care Center was responsible for quality control of this program. In order to ensure the quality of screening, experts from the Tianjin Women and Children Health-care Center paid frequent visits to screening centers in each district and county for data check-up, recheck and verification.

## Results

Among a population of 268989 (537978 eyes), 44 cases (75 eyes) of congenital cataract (13 unilateral cases and 31 bilateral cases) were found, accounting for 0.016‰ of the total. Of the 44 cases of congenital cataract, 33 (9 unilateral cases and 21 bilateral cases) were confirmed to be punctate cataract, and 5 cases (1 unilateral and 4 bilateral) were total cataract, of which 1 case was complicated with cerebral dysplasia. Posterior polar cataract were detected in 4 cases (1 unilateral case and 3 bilateral cases), anterior polar cataract in 2 cases (bilateral), nuclear cataract in 1 case (unilateral), perinuclear cataract in 1 case (unilateral), and congenital microphthalmia and small corneal with crytalline cloudiness in 1 case. The result has been showed in Figure 2 A.

Among the 44 positive cases, 13 babies (32 eyes) risk abnormal visual development and need timely treatment, which is 0.048‰ of the total screen population. 1 particular case of anterior polar cataract whose turbid shape is a symmetrical triangle with a side length of about 2.5 mm has been reported.

All 44 cases of congenital cataract were discovered in the first screening. No new cases were detected in the second and third time when the babies were 6 month and 1 year old. Among the 44 positive cases, only 7 cases (15.91%) had a family history of congenital cataract, the mothers of 4 ba-

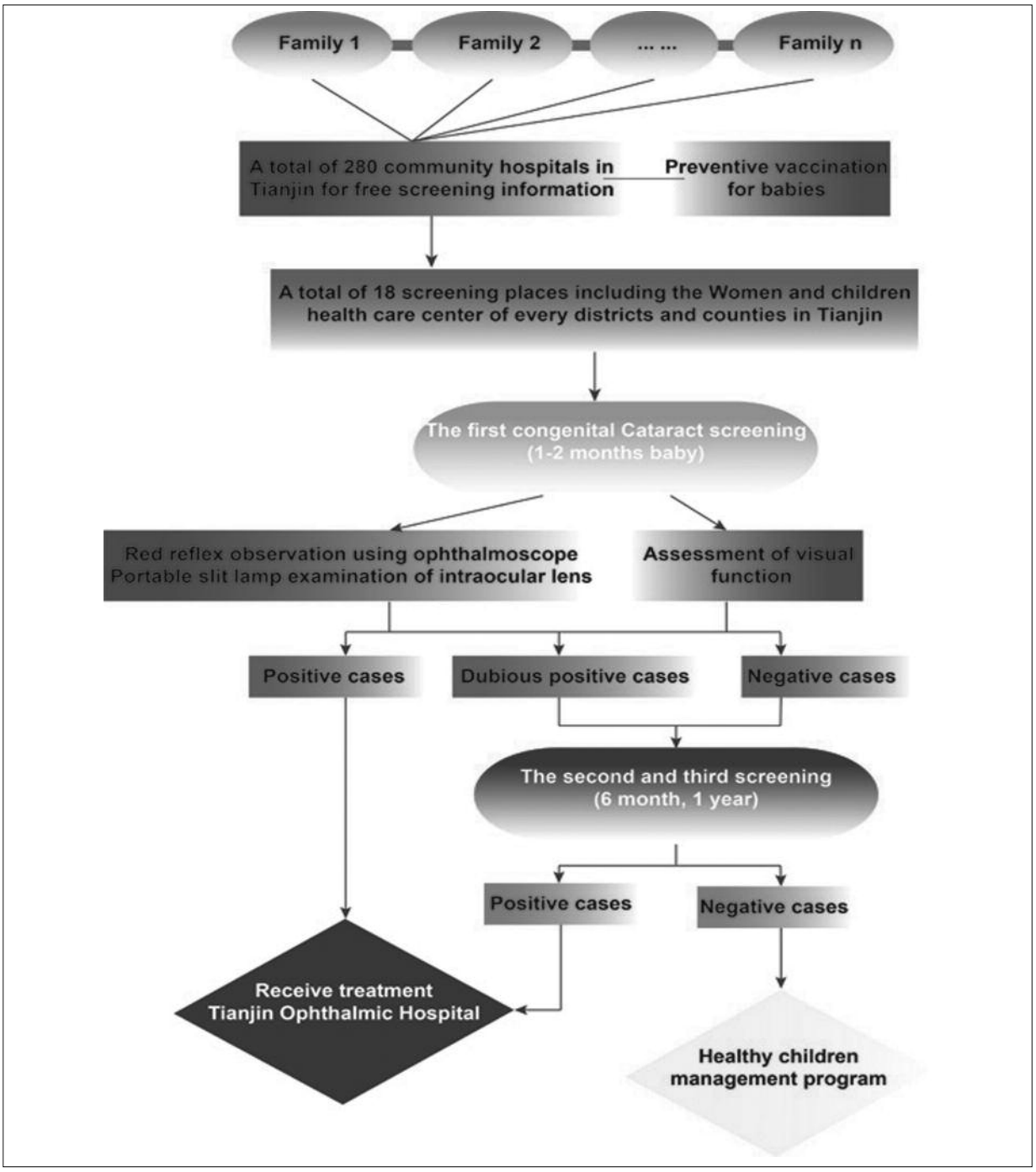


**Figure 1.** The screen procedures of the study.

bies (9.09%) suffered from in pregnancy, the mother of 1 baby (2.27%) had anemia complicated with gestational diabetes in pregnancy, the mothers of 2 babies aged 35 years or older (4.55%), another mother (2.27%) received X-ray of the chest in the 2nd months of pregnancy. And the remaining 29 cases (65.91%) can't be accounted for. The result has been showed in Figure 2 B.

## Discussion

It was reported that congenital cataract prevalence was 0.12‰-0.60‰[6,7] in some literatures and the incidence was 0.5‰-4‰ in a previous Chinese report[3,8,9]. In this study, we found 0.048‰ of all babies risk abnormal visual development and a prevalence rate of 0.16‰ for con-

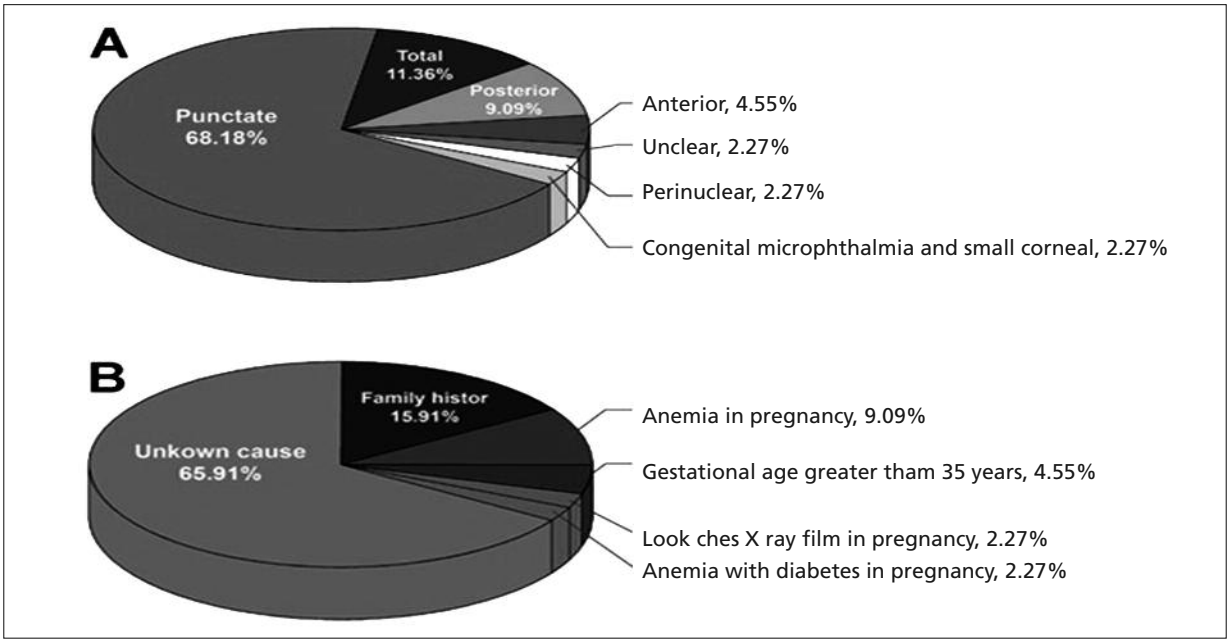


**Figure 2.** ***A,*** Distribution of types of congenital cataract in 44 cases. ***B,*** Distribution of types of risk factors for the 44 cases of congenital cataract.

genital cataract in Tianjin newborn babies from 2008 to 2011. This figure corresponded with international reports but was lower than that of other reports in China. It was possible that cases of congenital cataract were detected only in outpatients. Compared with this study, the age of the subjects, time of examination, and the methods in other screening programs were different, so that we had different conclusions.

The goal of this program is to discover infants with congenital cataract who risk abnormal visual development and to reduce visual disability in Tianjin babies. We need to adopt a kind of simple, more effective and quicker screen method that is generally acceptable to parents. As a result, we screened babies under normal pupil condition apart from those who have a family history of congenital cataract and need to go through mydriasis examination. Although peripheral opacity of lens may be undetected using this method, such as coronary cataract, partial perinuclear cataract, the possibility is low because babies' pupils expand in the dark. Moreover, we examined when the portable slit lamp and the eye were at the smallest angle so as to observe the periphery of lens clearly.

Congenital cataract has distinctive clinical and gene specificity, which may present different forms of lens opacity, such as coronary, punctate, perinuclear, nucleus, anterior polar, posterior pole, and total turbidity. The various types had different influence on the babies' eyesight and required varied treatment. In order to prevent amblyopia, babies suffering from posterior, perinuclear, nucleus, and total cataract which had an impact on the vision should be treated timely. Those had punctate, partial coronary cataract, and anterior polar cataract need to be followed for further observation. The 13 cases of congenital cataracts in this study were recommended with operation or mydriasis treatment according to their individual conditions. The remaining cases were observed in follow-up.

It is acknowledged that infants of two months old are in the key period of fixation reflex development. Operation in a later time will easily cause nystagmus, which is a symbol of vision recovery after cataract surgery. If nystagmus appeared before the operation, the postoperative visual acuity will have a satisfactory recovery. Otherwise, it is very difficult to restore normal eyesight or the eyesight may be even below 0.1.b Therefore, to screen infants for congenital cataract is crucial to prevention of amblyopia and reduction of visual disability. As is proved in this study, screening babies in normal pupil condition is a quick, simple, and acceptable way to find types of congenital cataract that effect on infants' visual development and especially those types occurred in the visual axis of babies.

The pathogenesis of congenital cataract is still unknown[5,10]. It could be familial or sporadic, and could be complicated with or without other ocular abnormalities or hereditary and systemic diseases.

Its occurrence was associated with internal and external factors. Exogenous factors include intrauterine (rubella, herpes virus, etc) infection in mothers of their early pregnancy (especially during 2 to 3 months of pregnancy), application of some drugs, exposure to X-ray, and metabolic diseases in mothers during pregnancy, such as diabetes, hypothyroidism, malnutrition and lack of vitamin. All these may affect fetal lens development and lead to congenital cataract.

## Conclusions

The results of this study showed that exogenous factors accounted for 18.18% of all congenital cataract cases and unknown factors for 65.91%. Endogenous factors are genetic factors, typically autosomal dominant inheritance. It is concluded that genetic factors accounted for only 15.91% of all cases, significantly lower than the findings of 20%-30% in previous relevant reports[4,11].

Congenital cataract was caused by genetic mutations and possessed significant genetic heterogeneity. However, whether the probability of gene mutation is associated with the different geographical location, dietary habits, and living and working environment of the parents or with maternal exposure to various external risk factors remained to be studied.

### Disclosure of Interest

The Authors declare no conflict of interest.

### Ethics

The study was approved by the Ethics Committee of Tianjin Disabled Persons' Federation (00012698-1).

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