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Screening for musculoskeletal system malformations and birth injuries in newborns: Results of a screening program in two hospitals in Shenzen, China

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ABSTRACT

Importance: There are a variety of musculoskeletal malformations and injuries that can occur in newborns. These can be a significant cause of perinatal death or a reason for miscarriage and can lead to long-term functional issues if not managed appropriately. There is no systematic and well-established screening program for neonatal musculoskeletal malformations and injuries in China now.

Objective: To report the incidence and types of congenital musculoskeletal malformations in two hospitals in Shenzhen City, to explore and discuss the details of the screening procedure and improve future prevention and treatment.

Methods: From October 2013 to May 2014, 2564 one-day-old newborns were screened by a pediatric orthopedic physical examination, in combination with ultrasonography when required, and the incidence and variety of diseases were recorded statistically.

Results: Among 2564 screened newborns, the following musculoskeletal conditions were identified: congenital muscular torticollis (CMT) (seven cases, 0.27%), hip subluxation (four cases, 0.16%), hip dysplasia (47 cases, 1.83%), congenital talipes equinovarus (CTEV) (two cases, 0.08%), congenital talipes calcaneovalgus (15 cases, 0.58%), polydactyly (nine cases, 0.35%), syndactyly (one case, 0.04%), and spinal hemivertebra (one case, 0.04%). Additionally, there were five (0.19%) neonates with birth injuries.

Interpretation: It is feasible to carry out neonatal screening and identification of musculoskeletal malformations and birth injuries in China. This is helpful as timely detection and early intervention for many of these conditions can avoid permanent functional impairment in these children.

KEYWORDS

Musculoskeletal system malformations, Newborn, Screening

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INTRODUCTION

Newborn screening for musculoskeletal malformations refers to health care screening in the newborn population for a number of conditions that jeopardize growth and development and cause disability. There are a wide variety of malformations of the musculoskeletal system in newborns, and some can be a significant cause of miscarriage, neonatal death, and infant death and functional impairment. The incidence of common musculoskeletal malformations in newborns varies according to different countries or ethnic factors, and it has been documented in recent years that the prevalence of musculoskeletal malformations is increasing year by year, about 3.8‰–9.69‰.[1–3] Prompt and effective intervention and treatment for many of these conditions will improve or normalize the child's limb function and avoid irreversible damage. At present, there is no systematic screening in our country for neonatal musculoskeletal system malformations and no international consensus regarding timing, methods, and criteria.

This study aims to report the incidence and types of malformations of the musculoskeletal system of newborns in two pediatric centers of Shenzhen City, to present details of the screening assessment in order to provide assistance for future efforts, so that more children can be treated promptly and early, to reduce disability and to improve the quality of life of the population.

METHODS

Ethical approval

This study was approved by the Ethics Committee of Shenzhen Children's Hospital, Peking University Shenzhen Hospital, and Shenzhen Maternity and Child Healthcare Hospital. The parents of each screened newborn gave informed consent allowing implementation of screening and disclosure of screening result information.

Participants

From October 2013 to May 2014, after excluding newborns with other severe diseases at birth, family refusal to undergo testing, and postnatal death, a single pediatric orthopedic surgeon performed clinical screening for musculoskeletal conditions in 2564 full-term newborns (gestational age above 37 weeks) born at Peking University Shenzhen Hospital and Shenzhen Maternity and Child Healthcare Hospital.

Diagnostic criteria

The diagnostic criteria were based on "Pediatric Surgery (4th edition),"[4] "Pediatric Clinical Surgery (5th edition),"[5] and "Tachdjian's Pediatric Orthopaedics (5th edition)."[6]

Data collection

The newborns who required screening for congenital musculoskeletal malformations were identified daily based on the 24-h delivery record of the obstetric department. The mother's name, admission number, gravidity, parity, length of gestation, method of delivery, volume of amniotic fluid, length of labor, and the neonate's name, gender, birth weight, and Apgar score were recorded. Physicians taught parents easy self-inspection practices while examining the newborns and recorded the physical findings. Families of newborns with negative results on initial examination were recommended to have regular health care physical examination for children, and those with clinical examination suggestive of congenital muscular torticollis (CMT) and/or developmental dysplasia of hip (DDH) were recommended to have neck and/or hip ultrasonography according to the Graf method. Newborns with abnormal ultrasound findings required prompt intervention and regular outpatient follow-up, those with normal ultrasound findings were advised to have regular routine health care physical examinations.

Physical examination methods

Each neonate was examined in the supine position in a room temperature maintained at 25°C.

The examiner is positioned at the head of the baby, placing the newborn in the supine position flat on the examination table with the head and neck draped and the neck mildly retroverted, allowing the neck to be exposed clearly. The examiner gently moves the child's head for passive rotatory range of motion and compares rotation bilaterally, assessing whether and to what extent neck rotation is limited. Bilateral neck symmetry, masses, erythema, or ecchymoses can also be observed. Palpation compares the sternocleidomastoid (SCM) muscles bilaterally for symmetry, with or without thickening, masses, and cords. A suspicious positive sign is indicated by limited rotational movement of the neck, thickening of SCM or palpation of suspicious masses, and cords (Figure 1).

The examiner moves to the feet of the baby, and first checks the neonatal face for asymmetry, checks the hands and feet for deformities, and examines the active and passive movement of the extremities in a quiet state. Next, removing the coat and pants to fully expose trunk and limbs, these are checked for deformities. Afterwards, the examiner can check passive and active upper and lower limb movements for movement disorders. Duplication of one or more digits in whole or part is diagnostic of polydactyly. Diagnosis of syndactyly malformation is confirmed by pathologic fusion of soft tissue or (and) skeletal structures of adjacent digits. Congenital talipes equinovarus (CTEV), also termed clubfoot, is diagnosed if there was forefoot adduction,

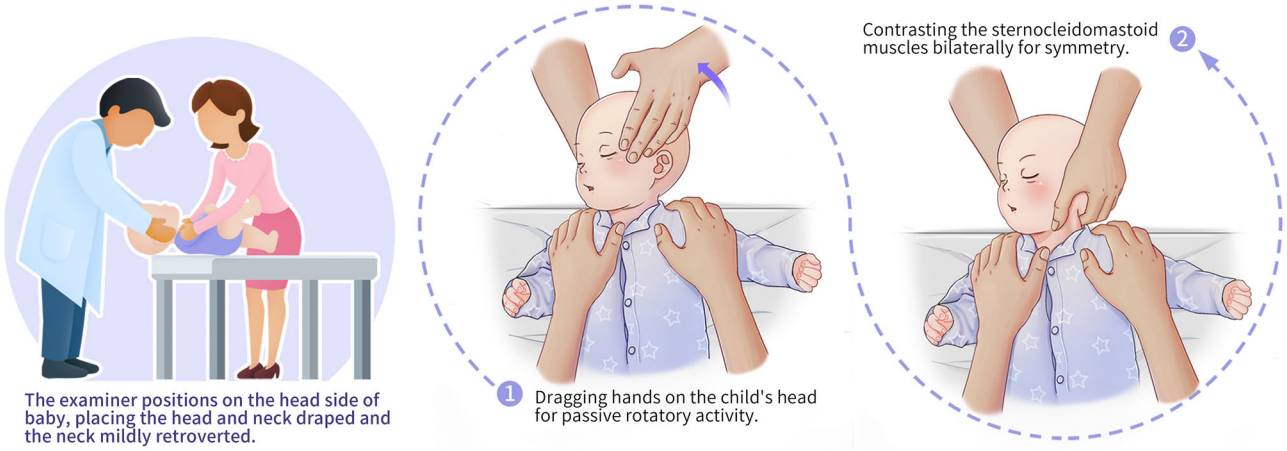

FIGURE 1 Physical examination methods for neck.

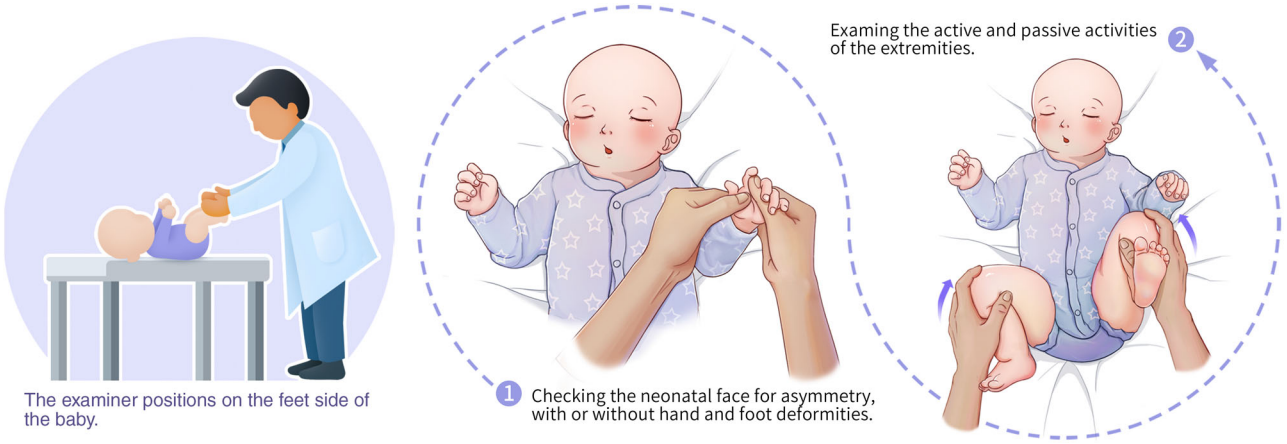

FIGURE 2 Examination of abnormal motion and malformation of limbs.

internal rotation, medial displacement of the midfoot, and high arched deformity, and the hindfoot was varus with tibial internal rotation, and external force could not correct those deformities (Figure 2).

The infant is dressed carefully to keep the upper body warm while fully exposing both lower extremities. The examiner checks for deformities of both legs on visual inspection, including asymmetry of skin folds of the thighs or buttocks, and limb discrepancy. The examiner places the infant's hips and knees in flexion with the feet held together, and examines the heights of both knees. If unequal knee heights were present, this is a positive Galeazzi/Allis sign. The examiner flexes the infant's hip and knee at 90° and allows gradual abduction of the hips to check for limitation of abduction. Abduction test is positive if the hip motion is less than 80°–90°. It is important to note that during gradual abduction of the hip, the Ortolani test is positive if there is a joint click/clunk and then the thigh could reach the examination table. Finally, with the infant's knees both in flexion, if the hip can be moved externally by thumb pressure and automatically relocates when the pressure is released, that is a positive Barlow test, which suggests hip instability (Figure 3). Finally, the examiner fully dressed the newborn.

Ultrasonography

Neonates with limited neck rotational movement, thickened or palpable SCM muscles with suspicious masses, and cords were evaluated by neck ultrasound for bilateral SCM muscle thickness, muscle morphology, and surrounding echogenicity. It is also necessary to observe whether a mass was present and to visualize muscle flow with color Doppler. If an infant younger than 6 months presents one sign of asymmetrical lower extremity skin folds, positive Allis sign, limited hip abduction, or positive Ortolani or Barlow test, the development and position of the femoral head and acetabulum were visualized and evaluated by Graf ultrasonography (α and β angle).

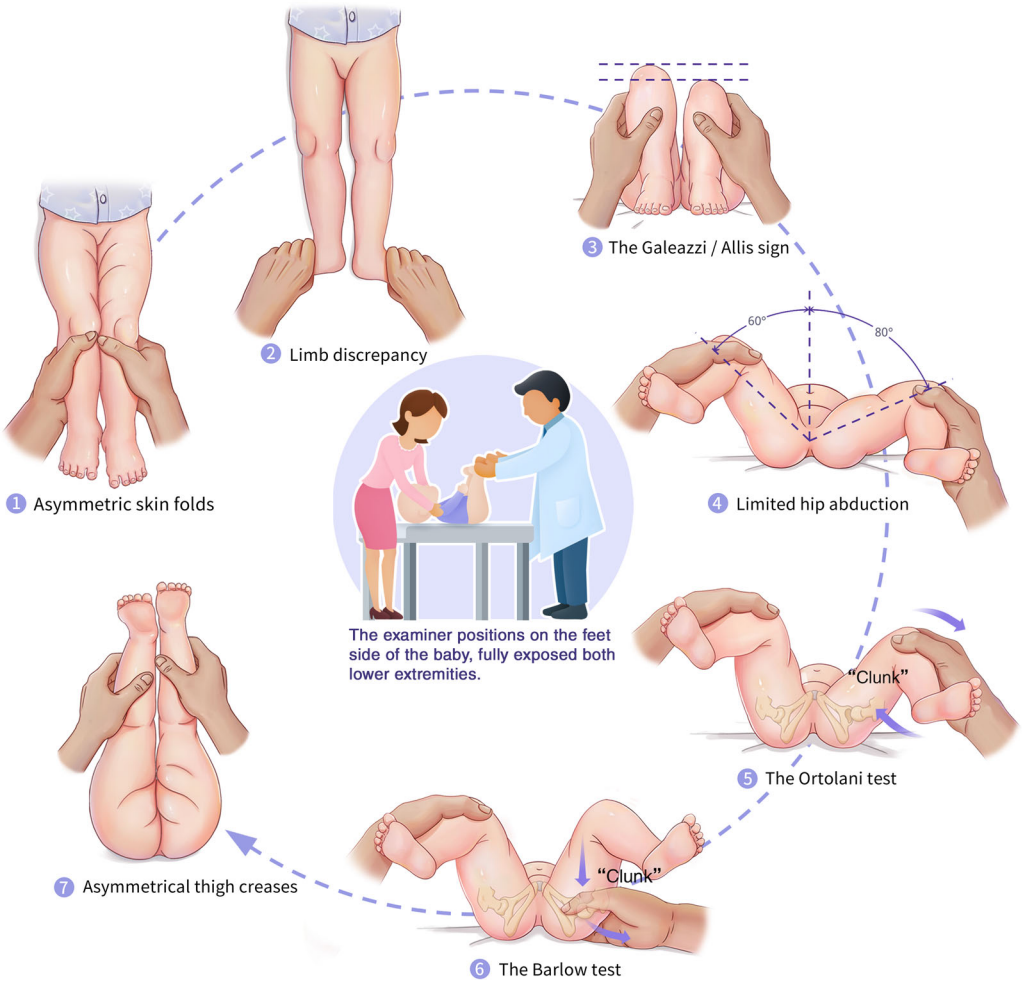

FIGURE 3 Physical examination methods for the hip.

Radiographic inspection

Newborns with limb swelling deformity, local tenderness, and limited mobility were evaluated with corresponding radiographs for bone pathology.

Statistical methods

Screening data were recorded and reviewed. Microsoft Excel was used to record and analyze the data.

RESULTS

A total of 2647 newborns were included during the screening period. Musculoskeletal system screening was performed in 2564 newborns who met the inclusion criteria. The following musculoskeletal conditions were identified: CMT (seven cases, 0.27%), hip subluxation (four cases, 0.16%), DDH (47 cases, 1.83%), CTEV (two cases, 0.08%), congenital talipes calcaneovalgus (15 cases, 0.58%), polydactyly (nine cases, 0.35%), syndactyly (one case, 0.03%), and spinal hemivertebra (one case, 0.03%). In addition, there were five (0.19%) cases of birth trauma, including three cases of clavicle fractures, one case of femur fracture, and one case of brachial plexus injury (Figure 4).

Of 2564 newborns examined on the first postnatal day, only one had limited cervical rotation. Neck ultrasound of 44 newborns with asymmetric facial and bilateral SCM muscle palpation showed that 36 had abnormal thickness of SCM bilaterally. Follow up of 2564 newborns revealed CMT in seven cases, of which only one presented at birth with facial asymmetry and bilateral asymmetry of SCM palpation in the neck.

A total of 90 suspected hip dysplasia cases were found by initial clinical examination, 27 (30%) were boys, 63 (70%) were girls, which showed twice as many girls as boys (Table 1). Those patients were separated by Graf classification subsequently, which showed that only 47 newborns were confirmed with type II DDH eventually, including 10 boys and 37 girls. There were two cases each of types III

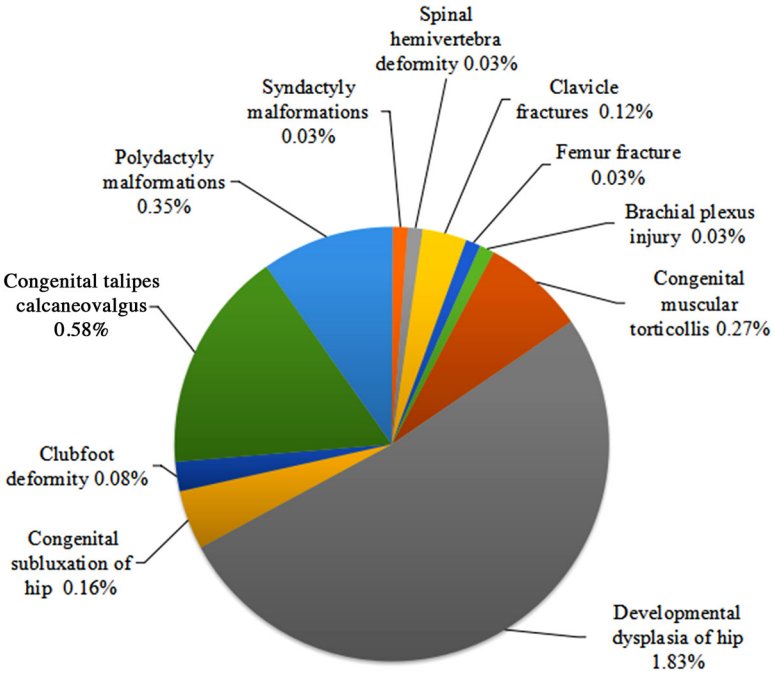

FIGURE 4 The screening results of musculoskeletal system malformations in 2564 newborns.

TABLE 1 Sex and Graf classification in newborns with suspected hip dysplasia by clinical examination

Graf classification	Gender, *n*	
	Male	Female
I	17	22
II	10	37
II	0	2
IV	0	2

and IV, all of whom were girls. Of the 10 boys with type II DDH, three were right, three were left, and four were bilateral. Of the 37 girls with Graf II, six were right, nine were left, and 22 were bilateral.

Physical examination of the hip joints revealed 121 neonates with asymmetric skin folds of the lower extremities, including 33 boys and 88 girls, 49 cesarean deliveries, and 72 vaginal deliveries. Ultrasound examination revealed Graf type I in 27 patients, type II in 32 patients, type III in two patients, and type IV in two patients. Of the three children with associated lower extremity disproportion, one was boy and two were girls, two were delivered by cesarean section, and one was delivered vaginally. Two were Graf type III and one was type IV. The abduction test was positive in 63 cases including 28 boys and 35 girls, 27 cesarean sections and 36 delivered vaginally. There were 12 ultrasonographically suggestive of Graf type I, 15 type II, 2 type III, and 2 type IV. Two cases had a positive Ortolani test and two had a positive Barlow test, all of whom were girls born by cesarean section, and the ultrasound found Graf type III in two and type IV in two (Table 2).

DISCUSSION

Routine newborn screening programs must be simple, noninvasive, effective, easy to implement, and less costly. Examiners can perform general physical examinations, but the key is to establish a standardized approach that can be implemented. A successful screening program should also include effective health education for parents, with brochures and pictures produced to preach all parents and increase their awareness. In addition, because some musculoskeletal malformations are developmentally dynamic, it is essential to refine a timely and effective follow-up procedure. Early diagnosis can not be established by physical examination alone. Positive signs in physical examination sometimes are not clinically significant, no conclusion can be drawn from the physical examination findings alone. The ultrasonography should also be combined. High-risk newborns need to be followed up and to ensure that they are diagnosed and treated early.

In this study, CMT was diagnosed in seven infants postnatally, accounting for 0.27% of the total newborn screening, lower than the incidence reported by other research (0.3%–2%).[7–10] In addition, there were 47 (1.83%) infants with DDH; four (0.16%) with hip subluxation and dislocation. The most common physical examination findings associated with ultrasonographic evidence of hip dysplasia were asymmetric skin folds and positive abduction test. Subluxation and dislocation of the hip can present with asymmetric

TABLE 2 Physical examination and ultrasound results of hip joints

Variable	Asymmetric skin folds	Lower limb discrepancy	Positive abduction test	Positive Ortolani test	Positive Balow test
Sex					
Male	33	1	28	0	0
Female	88	2	35	2	2
Delivery					
Cesarean section	49	2	27	2	2
Vaginal delivery	72	1	36	0	0
Graf classification					
I	27	0	12	0	0
II	32	0	15	0	0
III	2	2	2	2	0
IV	2	1	2	0	2

skin folds of the lower extremities, lower limb discrepancy, positive abduction test, positive Ortolani test, and Barlow test. In addition, we identified two (0.08%) cases of clubfoot, which has an incidence ranging from 0.1% to 0.3%.[11–13] As the population screened in the current study was newborns born in two hospitals in Shenzhen City over a short period of time, the incidence rates may not represent the overall incidence in the entire region.

It is obvious from this study that neonatal musculoskeletal system malformations can occur and can be detected early by a standardized physical examination. However, the special characteristics of hip development, as well as the limitations of the examiner's technical level, make our newborn screening for certain musculoskeletal system malformations difficult. DDH is a spectrum of disorders affecting the developing hip, ranging from slight loosening of the ligaments to complete dislocation.[14,15] The prognosis is unpredictable, and several studies are on how to screen for hip dysplasia early and which signs are more clinically meaningful.[14,16–21] Although skin fold asymmetry is not considered clinically significant by many physicians,[15] this sign has been described as one of the important suggestive findings of DDH.[16,17] Many reports recommend referral to a specialist hospital if the physical examination reveals positive signs such as skinfold asymmetry, leg length discrepancy, and hip click, but clinicians may not be well-trained to master these methods.[16,19,20,22–24] It has been documented that the presence of skinfold asymmetry is the most common reason for referral,[22] because it is relatively objective and convenient for general practitioners, pediatricians, and even parents. Ortolani and Barlow maneuvers require some training and experience, take time for pediatricians to learn, and may not be suitable for large-scale screening. From our study, the detection rate of DDH was significantly higher when positive signs were detected by physical examination, which are easier to perform in primary hospitals by pediatricians, and consideration of the combination of risk factors, including gender, family history, and breech presentation for DDH, form part of the initial evaluation in order to reduce missed cases.

As part of a child's orthopedic check-up, neonatal screening for musculoskeletal malformations is safe, effective, less costly than later assessment, and can be widely accepted. With appropriate configuration of medical resources, almost all maternity hospitals can perform a routine physical examination of newborns by obstetricians or neonatologists within 24 h after birth. This screening is based on established maternal and child health procedures, suitable newborn screening methods and should include an assessment of musculoskeletal malformations. We believe that every doctor or nurse who is specifically trained newborn examination methods, can perform an early evaluation of newborns for musculoskeletal system malformations, identify high-risk factors, and facilitate treatment and refer to a special hospital. Hence, this protocol plays an important role in early detection of the congenital disease. Also, through the newborn screening, we can detect birth injury timely, improving birth injury care and infant comfort, which can not only benefit the children but improve the doctor–patient relationship.

This study has provided viable evidence for the development of newborn screening and identification of musculoskeletal system malformations in China. First, a large proportion of parents support newborn screening and agree to participate in screening. Second, neonatal screening for musculoskeletal system malformations is technically feasible and the results are objective, simple, and rapid, which suitable for large-scale census. A well-established follow-up system will minimize false positives and false negatives at initial and subsequent test and will also enable early detection, timely treatment, and burden reduction of the

disease. Finally, health management can promote prevention and early detection and can improve health literacy of the population. Parents can be made aware of musculoskeletal discorders that can be prevented or require early treatment by providing information brochures in a range of healthcare setting, including prenatal units, obstetric clinic, antenatal diagnosis clinic, birth certificate issuing department, newborn parents' school, and pregnant women's school.

CONFLICT OF INTEREST

The authors have no conflicts of interest.

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