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*Original Article*

# Fetal chondrodysplasia punctata: a clinical study of five cases

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**Abstract:** Background: Chondrodysplasia punctata (CDP) is characterized by irregular calcification of the epiphyseal cartilage in the neonatal or infant period, which is determined by both genetic and non-genetic factors. Objective: To explore the prenatal ultrasonographic manifestations, genetic etiology, and pregnancy outcomes of fetal CDP. Methods: Five fetuses from three families diagnosed with CDP by prenatal ultrasound were retrospectively analyzed, and the clinical features, genetic testing results, and pregnancy outcomes of the CDP cases were analyzed. Results: Prenatal ultrasound showed strong punctate echogenicity of the long diaphyseal epiphysis (mainly in the femur, tibia, and humerus) in all five fetuses. Four fetuses (4/5, 80%) had short long bones. Three fetuses (3/5, 60%) had spinal anomalies and one (1/5, 20%) had Binder syndrome. Cataracts were present in two fetuses (2/5, 40%). Genetic testing was performed on all five fetuses. Chromosome karyotype and chromosomal microarray analysis showed no abnormalities. However, some region-specific pathogenic variants were identified in whole exome sequencing. Labor was induced in four fetuses. Only one fetus was delivered at 39 weeks of gestation, transferred to the Department of Pediatrics for respiratory support due to dyspnea after birth, and had a good prognosis with follow-up. Conclusion: Fetuses diagnosed with CDP mostly exhibit genetic abnormalities and adverse neonatal outcomes. Fetuses with punctate strong echogenicity of the long diaphyseal epiphysis and short long bones identified on ultrasound must be comprehensively screened for other systemic malformations, particularly spinal dysplasia and facial malformations. Genetic testing is recommended.

**Keywords:** Chondrodysplasia punctata, prenatal ultrasound, genetic testing, pregnancy outcome

## Introduction

Chondrodysplasia punctata (CDP) is a group of bone dysplasias characterized by irregular calcification of the epiphyseal cartilage in the neonatal or infant period, which is determined by both genetic and non-genetic factors. It is characterized by multiple spot-like calcifications in the soft tissues of long bones, joints, and vertebral bodies on radiography [1]. The incidence of CDP is extremely low, with a live birth incidence of 1/100,000 [2]. At present, there are few studies on fetal CDP, and most are case reports. Therefore, the aim of the present study was to improve prenatal diagnosis and provide scientific prognosis-related prenatal counseling by analyzing the ultrasound features, genetic etiology, and perinatal outcomes of five fetuses with CDP.

## Materials and methods

### *Subjects*

This retrospective study included clinical data from five fetuses with CDP (involving three pregnant women) diagnosed by prenatal ultrasound in a specialized hospital graded 3A, between November 2021 and November 2023.

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*Ultrasound measurements*

Ossification of the diaphysis of the extremities was observed, and fetal short long bones were defined as below the 5$^{th}$ percentile of the same gestational age [3]. The shape and ossification of the spine and vertebral canal were observed in sagittal, coronal, and transverse sections. The facial contour was assessed in the nasolabial coronal and midsagittal sections, whereas the upper and lower alveolar arcs were evaluated in the transverse section to assess the presence of Binder syndrome. Enhanced peripheral lens echo showing "bilateral sign" indicated potential cataract formation. Fetal CDP was comprehensively diagnosed based on these ultrasonographic findings [4].

*Genetic typing*

Based on genetic testing, CDP is classified into three subtypes: autosomal recessive genotype (limb root, rhizomelic chondrodysplasia punctata [RCDP]), X-linked recessive genotype chondrodysplasia punctata (CDPX1), and X-linked dominant genotype chondrodysplasia punctata (CDPX2) [5].

*Research methods*

We collected general information, prenatal ultrasound data, postnatal images, microarray analysis (CMA) data, whole exome sequencing (WES) data, pregnancy outcomes, and follow-up records for the five patients by reviewing the medical record system.

*Statistical analysis*

Descriptive statistical analyses were used to summarize the data in this study. Continuous variables were expressed as means ± standard deviation (SD). Categorical variables were presented as frequencies and percentages. Statistical Package for Social Science (SPSS) 29.0 was used for the statistical analysis.

*Ethical approval*

This study was conducted in accordance with the Declaration of Helsinki. The study protocol was reviewed and approved by the Ethics committee (No. 2024-079) of our hospital. Informed consent was obtained from all the participants.

## Results

*General data and ultrasound findings*

The data of five fetuses with CDP from three pregnant women were collected. The diagnosis was made through ultrasound screening during the second trimester (**Figure 1**). The general characteristics of the participants are presented in **Table 1**. There were three cases of CDP-affected fetuses in family 1. The mean age of the three women in this study was (27.20± 1.92) years, and the mean gestational age at ultrasonography-based diagnosis was (22.57± 3.23) weeks. One of the five pregnancies was conceived by in vitro fertilization. All fetuses presented with long diaphyseal epiphyses and strong punctate echoes on ultrasound, including four cases (4/5, 80%) in the metaphysis of the femoral shaft, two (2/5, 40%) in the metaphysis of the tibial shaft, and three (3/5, 60%) in the metaphysis of the humeral shaft. Of these five fetuses, four (4/5, 80%) exhibited short long bones; three (3/5, 60%) spinal dysplasia; one (1/5, 20%) Binder syndrome; two (2/5, 40%) cataracts; and one (1/5, 20%) syndactyly.

*Genetic testing results*

Among the five fetuses, three underwent amniocentesis, one underwent villus puncture, and one newborn received peripheral blood for genetic detection after birth. No abnormalities were found in chromosomal karyotyping or CMA in any of the cases, but genetic variations were found in some regions by WES (**Table 2** and **Figure 2D**, **2H**).

*Pregnancy outcomes*

Four fetuses were chosen for pregnancy termination (**Figure 2A-C**, **2E-G**). The gestational period for termination was (24.11±5.74) weeks. Only the fifth fetus continued to full term pregnancy: a male infant was delivered vaginally at 39 weeks of gestation. The newborn was transferred to the pediatric department for respiratory support because of dyspnea after birth. The surviving infant grew well, except for a low nose bridge (**Table 2**).

## Discussion

Chondrodysplasia punctata was first reported in 1914 [6] and frequently entails multiple

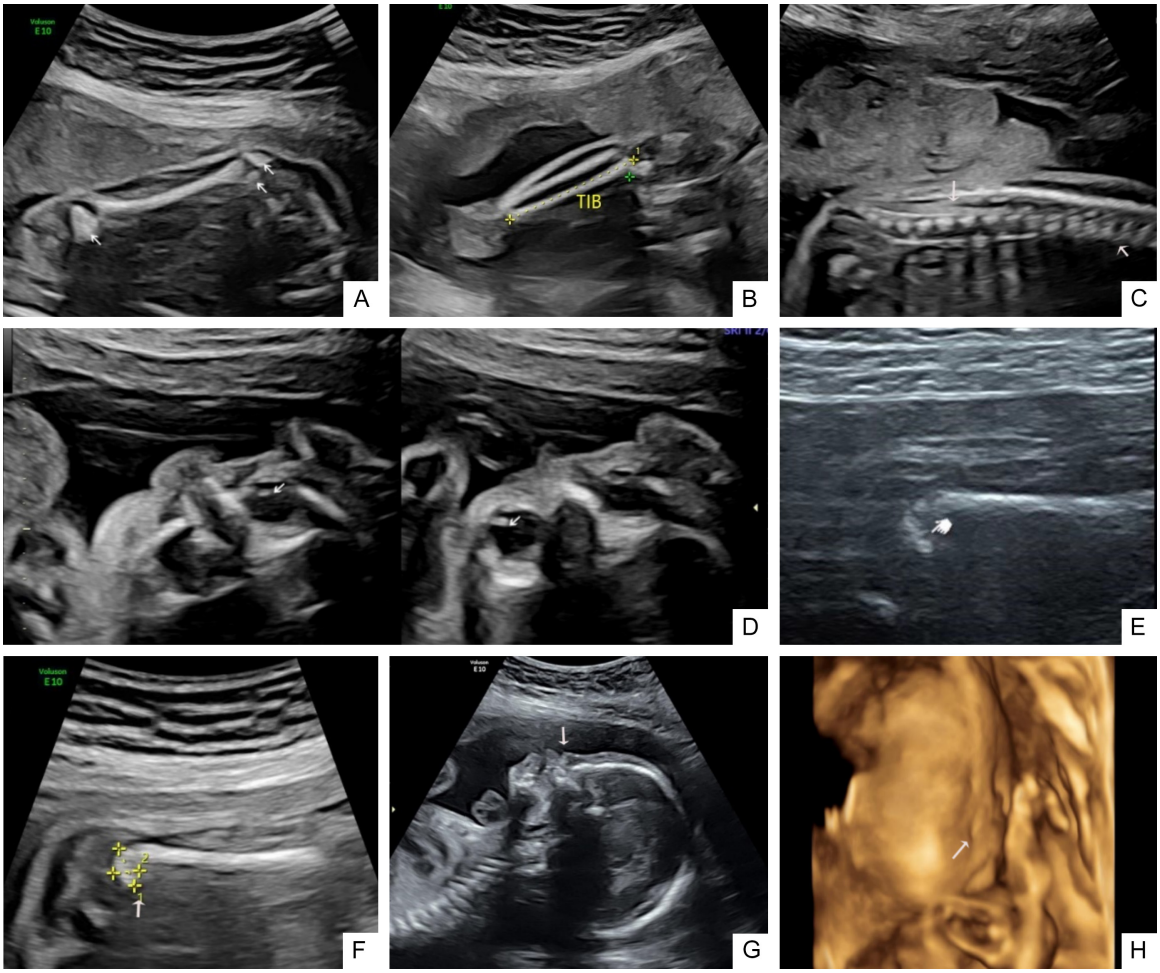


**Figure 1.** Ultrasound features in three fetuses. A-D. Fetus 1(24+6wg*). E, F. Fetus 4(24+6wg*). G, H. Fetus 5(24+6wg*). A, B. Epiphyseal stippling, thick metaphysis, shortening of the tibial and the femur; C. Small spine of the T5-11 vertebrae, coronal fissures in the T12, L1, and L2 vertebrae, and lowering conus medullaris; D. Enhancing bilateral peripheral echoes of the crystalline lens, showing "bilateral sign"; E. Epiphyseal stippling of the femur; F. Thick metaphysis of the femur; G. Flat nose bridge, collapsed nasal wings, nose tip lower than upper lip and short nasal bone; H. Three dimensional imaging collapsed nose deformity. Abbreviations: wg*, weeks of gestation.

organ damage, abnormal limb development, joint malformations, skin lesions, congenital cataracts, defects in the epiphyseal or cardiovascular system, and particular features (such as bridge collapse of the nose, protrusion of the forehead, high palate, or palate cleft, etc.). Therefore, multidisciplinary diagnosis, treatment, and symptom management are required. Prenatal ultrasonography of fetal CDP is highly valuable for diagnosis.

A literature search of PubMed revealed that prenatal ultrasound diagnosis of CDP is relatively uncommon, with most cases reported as individual case studies. The publication time was early, and the clinical and imaging data were incomplete. Fetal CDP ultrasonography should focus on observing ossification of the diaphysis of the long limbs. If a punctate strong echo is observed in the long diaphysis, further observation should be conducted on the combination of short long bones, spinal dysplasia, Binder syndrome, and cataracts [7, 8]. Based on the literature review [4, 6, 8-17], among the 22 reported fetuses with CDP, 20 exhibited shortened long bones (primarily humerus and femur); 19 had epiphyseal calcification; 11 had vertebral abnormalities (such as coronal clefts, flat vertebral bodies, hemivertebrae); 1 had cataracts; and 5 had Binder syndrome. In this study, punctate hyperechoic features were observed in all five fetuses, with the most common complication being short limb deformity and the other major systemic abnormalities in

**Table 1.** General fetal conditions and ultrasonic features of the 5 CDP cases

| Family (n=3) | 1 (fetus 1) | 1 (fetus 2) | 1 (fetus 3) | 2 (fetus 4) | 3 (fetus 5) |
|---|---|---|---|---|---|
| Maternal age (years) | 26 | 27 | 28 | 30 | 25 |
| Mode of conception | Spontaneous conception | Spontaneous conception | In Vitro Fertilization | Spontaneous conception | Spontaneous conception |
| Gestational age at diagnosis (wg*) | 24+6 | 20+2 | 18 | 24+6 | 24+6 |
| Strong echo dots of long diaphyseal | Bilateral humerus, femur, tibia | Bilateral humerus and femur | Bilateral humerus and femur | Bilateral femur | Bilateral tibia (Weak strong echo) |
| Shortening of the long bones | femur (3.5 cm, 0th)<br>humerus (2.2 cm, 0th) | femur (2.7 cm, 0.4th)<br>humerus (1.7 cm, 0th)<br>radius (2.2 cm, 0.7th)<br>ulna (2.5 cm, 1th)<br>fibula (2.3 cm, 0.4th) | femur (2.2 cm, 2.1th)<br>humerus (1.7 cm, 0th)<br>ulna (1.84 cm, 0.4th) | femur (3.6 cm, 0th)<br>humerus (3.4 cm, 0th)<br>radius (3.1 cm, 1.3th)<br>ulna (3.3 cm, 0.1th)<br>tibia (3.6 cm, 2.2th)<br>fibula (3.3 cm, 0.2th) | - |
| Abnormal spine | small thoracic 5- thoracic 11 vertebrae, coronal fissure in thoracic 12- lumbar 2 vertebrae, and low spinal conus | low spinal conus | low spinal conus | - | - |
| Binder syndrome | - | - | - | - | flat nasal bridge, collapsed nasal wings, and short nasal bone of (0.3 cm) |
| Others | Cataracts, syndactyly | Cataracts | - | - | - |

Abbreviations: CDP, chondrodysplasia punctata; wg*, weeks of gestation.

**Table 2.** Fetal genetic etiology and pregnancy outcome of the 5 cases of CDP

| Family (n=3) | chromosome karyotype | CMA | WES | CDP subtypes | Parental origin and phenotype | Pregnancy outcomes and gestational age at termination (wg*) |
|---|---|---|---|---|---|---|
| 1 (fetus 1-3) | Normal | Normal | PEX7 gene<br>c.121G>C (p.G41R)<br>c.337G>A (p.E113K)<br>complex heterozygous variation | RCDP | PEX7 gene: Paternal and maternal (normal phenotype) | Induction of labor (25+5, 20+6, 18+3) |
| 2 (fetus 4) | Normal | Normal | EBP gene<br>c.303G>A (p.w101)<br>heterozygous variation | CDPX2 | New mutation | Induction of labor (31+3) |
| 3 (fetus 5) | Normal | Normal | ARSE gene<br>c.1445C>A (p.P482H)<br>hemizygote variation | CDPX1 | ARSE gene: Maternal (normal phenotype) | Continue pregnancy, foot menstrual vaginal delivery (39) |

Abbreviations: CDP, chondrodysplasia punctata; CMA, chromosomal microarray analysis; WES, whole exome sequencing; RCDP, rhizomelic chondrodysplasia punctata; CDPX1, X-linked recessive genotype chondrodysplasia punctata; CDPX2, X-linked dominant genotype chondrodysplasia punctata; wg*, weeks of gestation.

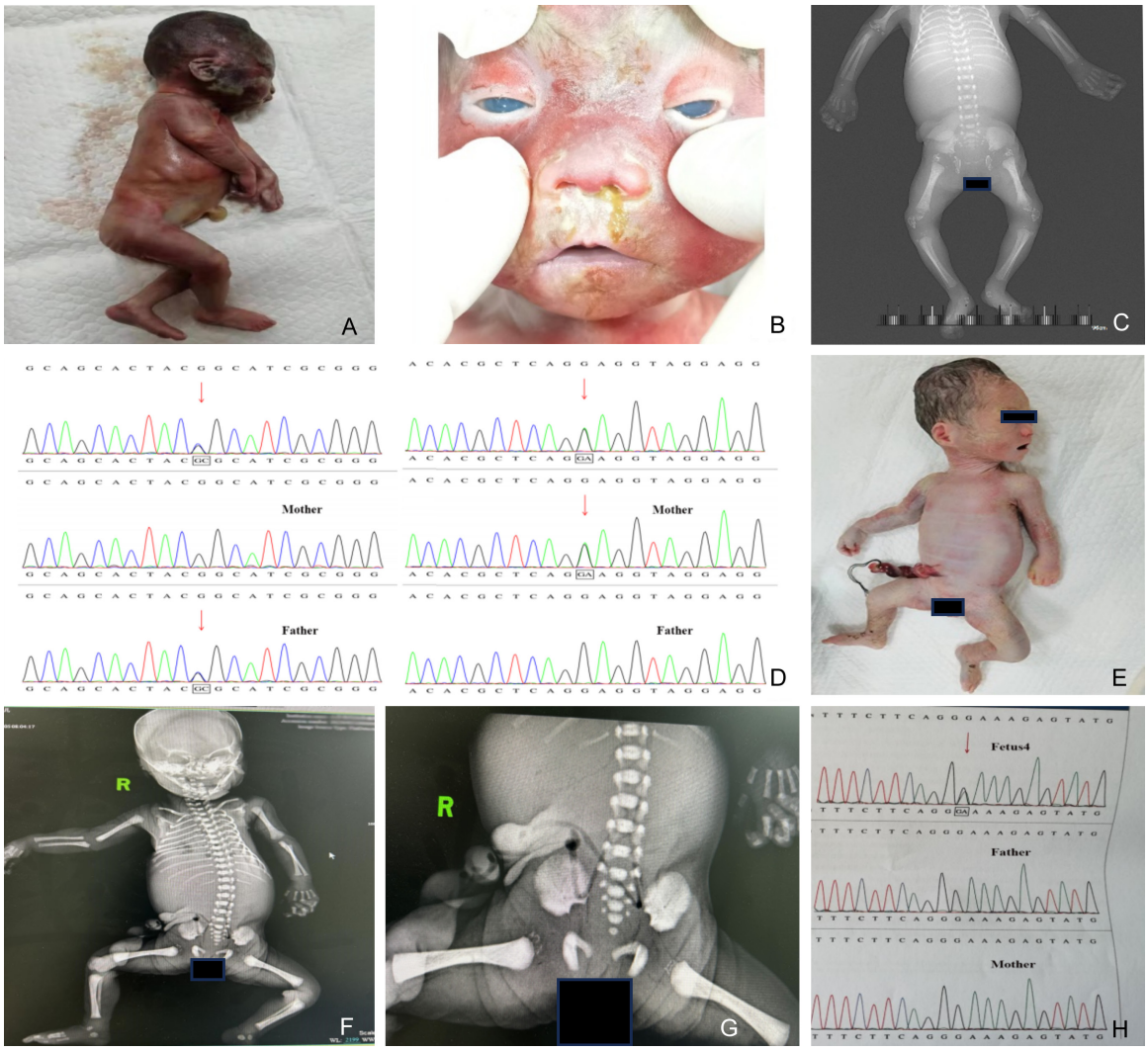


**Figure 2.** Clinical findings, radiological features and WES in two fetuses. A-D. Fetus 1(25+5wg*). E-H. Fetus 4(31+3wg*). A. Shortened upper arms and thighs; B. Flat nasal bridge and cataracts in both eyes; C. Multiple punctate ossification at the epiphyseal end of long bones and incomplete ossification of thoracic vertebrae by X-rays; D. PEX7 gene c.121G>C (p.G41R) and c.337G>A (p.E113K) exhibiting compound heterozygous variation, inherited from its father and mother; E. Shortened long bones of the limbs and flat nasal bridge; F, G. Shortened long bones and thick metaphysis of the femur with epiphyseal stippling by X-rays; H. EBP gene c.303G>A (p.W101) exhibiting a new mutation, no inherited from its parents. Abbreviations: WES, whole exome sequencing; wg*, weeks of gestation.

sequence being spinal dysplasia, cataracts, and Binder syndrome, which is consistent with previous studies.

In terms of etiology, fetal CDP exhibits heterogeneity and has been found to be associated with chromosomal aneuploidy disorders (such as trisomy 18 syndrome, trisomy 21 syndrome), monogenic disorders (such as CDPX2, CDPX1, RCDP, Zellweger syndrome, GM1 gangliosis, Smith-Lemli-OPitz syndrome, etc.), genetic disorders of vitamin K metabolism, maternal autoimmune diseases, as well as drug and teratogenic exposures (such as warfarin, phenytoin, alcohol, etc.) [18-20]. In our study, the three pregnant women denied a history of medication, teratogenic exposure, genetic disorders related to vitamin K metabolism, or autoimmune diseases. All five fetuses underwent genetic testing. No abnormalities were found in the chromosome karyotype analysis and CMA, whereas pathogenic variations were found in WES, which were considered RCDP, CDPX1, and CDPX2 subtypes.

According to the pathogenic genes, RCDP can be divided into the following five subtypes: RCDP1 related to PEX7; RCDP2, GNPAT; RCDP3, AGPS; RCDP4, FAR1; and, RCDP5, PEX5 [21-23]. In Family 1, three fetuses were found to have compound heterozygous variations in the PEX7 gene, c.121G>C (p.G41R) and c.337G>A (p.E113K), inherited from their father and mother, respectively. PEX7 is the only known pathogenic gene of the RCDP1 type, which encodes the peroxisomal stromal enzyme receptor [21, 24]. The clinical manifestations are proximal symmetric shortening of the limbs, joint contracture, cataracts, and various degrees of nervous system development retardation; the genetic mode is autosomal recessive [18]. The ultrasonographic findings of the three fetuses in this family showed that the epiphyseal end of the long shaft was thick and had multiple ossifications, combined with short limb deformity, coronal fissure of the vertebral body, and binocular cataract, which were highly consistent with the disease. Therefore, we considered that the RCDP1 subtype was caused by complex heterozygous mutation of the PEX7 gene. Finally, the three pregnancies were abandoned.

A heterozygous variant of the EBP gene, c.303G>A (p.W101), a pathogenic variant associated with CDPX2, was identified in the fetus of family 2. This was confirmed to be a new mutation that was not carried by the parents. The inheritance mode was X-linked dominant inheritance, with an incidence rate of approximately 1/100,000 [25]. CDPX2 is often characterized by male lethality and female morbidity, with typical clinical features, including ichthyosis, punctate cartilage, poor fertility, special facial features (cataracts, saddle nose, and other craniofacial defects), asymmetric limb shortening, and short stature. In this case, the female fetus presented with evident short-limb deformities, an enlarged femur epiphysis, and a flattened nasal bridge. We considered that the CDPX2 subtype was caused by this new mutation of the EBP gene.

In family 3, the mid-pregnancy ultrasound showed abnormal development in the middle of the fetus's face. Genetic testing of the amniotic fluid puncture revealed no abnormalities in the chromosome karyotype analysis or CMA. The pregnancy was continued, and a male infant with a flat nasal bridge was delivered at term. Due to breathing difficulties, the fetus was transferred to the pediatric department for continuous positive pressure-assisted ventilation, and the symptoms improved. Follow-up history revealed that the child's maternal family had two male relatives with flat nose bridges, one of whom died due to breathing difficulties. Further investigation identified a hemizygous ARSE variant, c.1445C>A (p.P482H), through WES analysis of peripheral blood, which was not present in the child's father but was found to be heterozygous in the mother and associated with CDPX1 without any phenotypic effects. CDPX1 is a relatively rare X-linked recessive congenital disorder affecting bone and cartilage development, and it primarily affects males [26]. The main manifestations include punctate calcification of the epiphysis, short stature, microcephaly, cataracts, developmental retardation, and nasal hypoplasia. In the present case, the newborn had weak strong echoes at the epiphyseal end of the tibia, accompanied by a flattened nasal bridge, which was consistent with the CDPX1 phenotype. Therefore, the heterozygous ARSE gene variation was considered to be a pathogenic variation.

CDP can be classified according to genetic testing, and prognosis varies greatly among the different types. RCDP has been confirmed to be the most widely understood and worst prognosis type, often accompanied by severe mental retardation, and most children die due to respiratory failure. Most children with other CDP types survive for an extended period but typically have short stature in adulthood. The majority of CDPX1 fetuses do not exhibit obvious major organ structural abnormalities during pregnancy, making prenatal diagnosis challenging and prone to oversight. The medical history and genetic study of fetus 5 in this cohort offers a novel diagnostic approach for fetal facial dysplasia identified during prenatal examination. This subtype generally has a favorable prognosis, normal intelligence, and standard life expectancy. In conclusion, a thorough and accurate medical history, ultrasound imaging features of intrauterine fetuses, and WES types provides crucial guidance for comprehensively assessing the long-term prognosis of fetuses.

Binder syndrome and CDP have a strong correlation [27]. Binder syndrome is a midline facial developmental disorder associated with the absence of the nasal spine, resulting in a small nose tip, flat nose bridge, short nose column, acute change in the naso-lip angle, raised upper lip, and a tendency towards a tertiary occluding malocclusion [28, 29]. In a few types of CDP, long bones exhibit normal length and structure, and punctiform ossification on ultrasound findings may be subtle, leading to potential oversight during diagnosis [10]. Therefore, identifying Binder syndromes can offer valuable clues for diagnosing CDP. In this study, fetus 5 was diagnosed with Binder syndrome through ultrasonography; it exhibited both weak and strong echoes in the bilateral tibial metaphyses. CDPX1 type was confirmed using WES after delivery.

### Limitations of the study

The sample size was small, and termination was primarily chosen for cases with ultrasound diagnosis combined with multiple malformations, resulting in a limited number of follow-up cases for surviving newborns with CDP. Subsequent studies should aim to expand the sample size and include additional genetic testing.

In summary, when prenatal ultrasonography reveals a long diaphyseal point with a strong echo combined with short long bones, comprehensive screening for other systemic malformations should be conducted, paying particular attention to spinal dysplasia and facial malformations. Genetic testing is recommended for early detection of genetic abnormalities and definitive typing for fetuses with a high suspicion of CDP. Considering the different prognoses of the various subtypes of CDP, we aimed to provide accurate prenatal consultations to patients, guide fertility-related decision-making, and offer guidance for family reproduction planning.

### Disclosure of conflict of interest

None.

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