# Two Case Reports of Chondrodysplasia Punctata Diagnosed by Prenatal Ultrasound Screening.

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**Abstract**

The Binder phenotype is characterized by hypoplasia of the nasal bone and maxilla. It is one of the important ultrasound findings when screening for osteochondrodysplasia or chromosomal abnormality.

We have experienced two cases of the Binder phenotype detected by fetal ultrasound screening in the second trimester, and the patients were diagnosed postnatally with chondrodysplasia punctata (CDP). The pathogenesis was considered genetic arylsulfatase E deficiency and vitamin K deficiency caused by severe hyperemesis gravidarum. The measurement of the frontonasal angle is useful when screening for a Binder phenotype, and proximal femoral epiphysis calcification is critical to CDP diagnosis. Since the screening of the facial phenotype is crucial to point out CDP, which is often complicated by cervical spondylosis or respiratory stenosis, careful consideration is necessary when CDP is suspected based on a Binder phenotype detected by fetal ultrasonography.

**Key words: chondrodysplasia punctata, Binder Phenotype, frontonasal angle, *CDPX1*, arylsulfatase E deficiency, hyperemesis gravidarum**