عنوان مقاله:
دیسپلازی تاناتوفوریک در نوزادان دوقلو

محل انتشار:
مجله علمی ناباروری ایران, دوره 3, شماره 1 (سال: 1391)

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خلاصه مقاله:
Thanatophoricdysplasia (TD) isanosteochondrodysplasiaalwayslethalintheneonatalperiod. Thevastmajorityofcasesareduetodenovomutations. Itisdividedintotwotypes: ashortcurvedfemurcharacterizestype 1, whileastraighterfemurwithcloverleafskullcharacterizestype 2. Inthanatophoricdysplasiathelimbsareveryshort. Theribcageissmall. Thevertebralbodiesofthespinearegreatlyreducedinheightwithwidespacesbetweenthem. Autosomaldominantmutationsinthefibroblastgrowthfactorreceptor 3 gene (FGFR3), whichhasbeenmappedtochromosomeband 4p16.3, resultsinbothsubtypes. Thisconditionhascharacteristicsonographicfeaturesthatsuggestthediagnosisprenatally. Thanatophoricfetusesusuallydiewithinthefirst 48 hoursoflifefrompulmonaryhypoplasiacausedbyanarrowthorax, leadingtorespiratoryinsufficiency. Wereportedtwindizygotecasesoftype 1 .TDwithsimilarfindingsadjustingwithTDforthefirsttime, alongwithashortreviewoftheavailableliterature

کلمات کلیدی:
Thanatophoric dysplasia, Newborn, Twins