INTRODUCTION
The global prevalence of skeletal dysplasias worldwide has been seen of 2.4 cases of 10,000 births and the prevalence of lethal dysplasia 0.95-1.5 cases of 10,000 births. In general, they are rare diseases that become 5% of the genetic disorders identified during the neonatal period.

The three most common skeletal dysplasias are: Thanatophoric Dysplasia, Osteogenesis imperfecta type 2 and Achondrogenesis. Skeletal dysplasias can be identified in sonographic study early because the vast majority of the bones have an early ossification.

OBJECTIVE
Review of one of the three most common skeletal dysplasias through the presentation of a case diagnosed in our Healthcare Unit and thus maintain and active behavior in the detection and management of these diseases.

METHODS
We present a case of Thanatophoric Dysplasia and a review of the three most common skeletal dysplasias. Finally, we examine the best diagnostic method and the most important lethal signs that are useful for management of the case and counseling the family.

CONCLUSION
The skeletal dysplasias are rare diseases of the genetic disorders identified during the neonatal period but physicians should be actualized in its diagnosis and management.

The primary method of screening and study is the ultrasound. Additionally a definitive diagnosis is possible using cell-free fetal DNA in maternal plasma.

It is important to know the most predictive signs and identify them in the ultrasound study. The most reliable sign is the calvarium demineralization.

Identify the lethality of the disease can change the treatment and management of the case.

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