

# Atelosteogenesis Type II :

## case report Comparison between prenatal ultrasound, fetal autopsy and X-Ray findings

Hammami A, Zeghal D, Slimani S, Kdous S, Kehila M, Mahjoub S, Ben Hmid R, Channoufi MB  
Center Of Gynecology And Obstetrics if Tunis (CMNT), Department "C", Tunisia

### Introduction :

Mc Alister's syndrome also called "atélostéogénèse type II" is an autosomal recessive disease. It is a very rare perinatal dysplasia (prevalence 1/50000) characterized by a shortening of the limbs, a dysmorphic syndrome and radiological abnormalities. The diagnosis can be made based on radiological examination of the entire skeleton.

We report the case of Mc Alister's syndrome suspected by prenatal ultrasound and confirmed by autopsy.

### Objective :

Insist on the importance of knowing Mc Alister Syndrome for better genetic counseling.

### Patients and methods:

Comparison between prenatal ultrasound findings, fetal autopsy outcome and X-Ray of the fetal's entire body.

### Case report :

we report the case of a patient of 31 years, blood group B positive, married for 5 years, first-degree consanguineous marriage, G3P2, with a history of termination of pregnancy in the 2nd quarter during her first pregnancy for abnormalities of limbs (unlabeled malformation) and a living 3years apparently healthy child. The patient was referred to our department at 24GW for limb anomalies discovered during morphological fetal ultrasound.

**Prenatal ultrasound** showed shortening of the long bones (fig 1,2,3) (below the 3rd percentile), club feet, without other associated anomalies. Mc Alister Syndrome has been suspected for consanguinity and recurrence of the malformation detected on ultrasound .



Fig 3 Cerebellar diameter =23GW



Fig 1 femur length

Fig 2 humerus length

A termination of pregnancy was decided after a written consent of the patient.. **Fetal autopsy** found short limbs, deformation of the fingers and facial dysmorphism (fig4,5,6,7).



Fig 4, 5: short limb



Fig 6: hand deformation

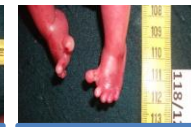


Fig 7: feet deformation



Fig 8: X-Ray of the entire body

**X-Ray of the entire body** of the fetus (fig8) confirmed the diagnosis by showing short, massive, with widened metaphyses. Epiphyses of the knee were irregular and flattened. The spine and pelvis were however slightly modified.

### Discussion:

What was missing in our investigation is the histological studies that could have shown the lack of cartilage matrix, manifested by increased chondrocyte density for age, cells forms somewhat limited and various gaps in the inter-cellular matrix.

### conclusion :

Mac Alister syndrome is a rare malformation, autosomal recessive; we should think about it each time we find short limbs, especially if there is a precedent case in the family.