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CAUDAL REGRESSION SYNDROME: A COMPELLING PICTURE TO CALL FOR MATERNAL DIABETES PREVENTION, SCREENING AND MANAGEMENT DURING PREGNANCY

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First described by French surgeon and anatomist Bernard Duhamel in the late fifties, caudal regression syndrome refers to a heterogenous group of congenital disorders that affect with various degrees the caudal part of the spine, the spinal cord, the urogenital system and the lower limbs [1]. Cases have been reported of associated cleft palate and congenital heart diseases and prognosis of the condition depends on the severity of organs and functions affected. Caudal regression syndrome is extremely rare and sporadic, and results from neurulation defects during the first 28 days of embryologic development [3]. Its pathogeny is multi-factorial and identified etiologies include uncontrolled maternal diabetes, vascular hypoperfusion-induced oxydative stress and genetic predisposition, through the mutation of gene VALGL1 [4,5]. Prevalence is estimated around 1/20 000 and diabetes bears the brunt of a consistent part, with nearly 20 to 25% of mothers of children with caudal regression syndrome reporting uncontrolled diabetes [6].

We report the case of a 34 years secundigraviade, with documented family history of diabetes, who had a first cesarean section for breech presentation over narrow pelvic outlet . The newborn weighed 3700gr, had an Apgar score of 10 at 5 minutes and presented no abnomality at pediatric check up. The post-partum was uneventful for the patient and the newborn. The second pregnancy was unplanned and she consulted for her first prenatal care check up at 33 weeks of gestational age. A 75gr oral glucose tolerance test was performed given her family history of diabetes and BMI at 29 and revealed 2 abnormal values out of the 3. She was put under strict diet and insulin therapy. Fetal biometric parameters at ultrasound scan showed a consistent difference between an abdominal circumference above 90% centile for gestational age and a femur length at 10% centile only. A hydramnios was identified but given the examination conditions and patient's BMI, further fetal morphological analysis proved difficult. The patient delivered at 38 weeks of gestational age through a planned cesarian section for scarred uterus over narrow pelvic outlet. The newborn was a girl and weighted 3950gr for a length of 37 cm. APGAR score was 10 at 5 minutes. Morphological examination revealed short bowed lower limbs (Figure 1), a perforated anal orifice and well-individualized female external genitalia (**Figure 2, 3**).

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Figure 1: Morphological aspect

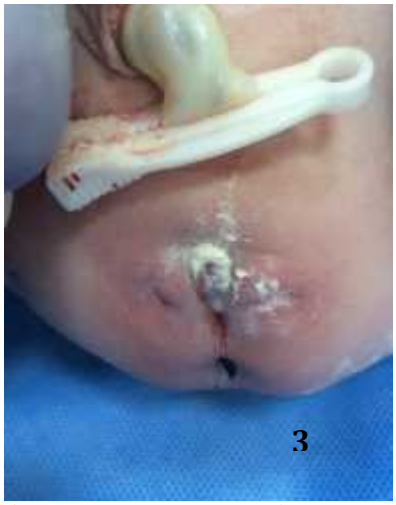

Figure 2: Aspect of lower abdomen and limbs; **Figure 3:** Aspect of external genitalia and anal orifice

A CT scan was performed and revealed a total agenesis of the coccyx and partial agenesis of the sacrum. No spinal cord or urogenital abnormalities were found. Heart ultrasound identified a mild hypertrophic cardiomyopathy. The newborn was referred to the Cardiology Department for management and surveillance and Children Traumatology and Orthopedics department for functional management of the lower limbs impairment.

Caudal regression syndrome is rare but the quality of life and heavy management of associated abnormalities and impairments after the child is born highlights the problem of diabetes management among women in reproductive age and the crucial need to standardize and generalize preconceptional care for this category of patients in order to control diabetes and decide the optimal timing for conception. This is particularly important in Morocco, where diabetes is reaching endemic levels and where prevention, screening and management programs should span through a multidisciplinary and comprehensive approach, and include a strong sexual and reproductive health component for women.

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