A case of vulvar mass

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Objective

Only a few cases of prenatally diagnosed congenital genital/perineal masses have been reported. Differential diagnosis includes a great variety of anomalies. Frequently it isn’t an isolated finding and prognosis depends on associated anomalies. We report a case of prenatally diagnosed congenital vulvar mass which was associated to a LUMBAR syndrome.

Methods

A 29-year-old woman presented for routine midtrimester ultrasound at 20 weeks’ gestation. Previous pregnancy tests were completely normal. At genital scan, a female external genitalia were identified based on the visualization of four parallel lines representing the labia majora and minora. A mass between both labia minora was visualized. A detailed anatomy scan of the fetus didn’t reveal any other anomaly except for a single umbilical artery. An amniocentesis was offered to the parents which revealed 46,XX karyotype, normal microarray and a negative adrenal hyperplasia study in accordance with the phenotypic sex. Our suspected diagnosis was fetal transient cliteromegaly versus fetal vulvar tumor. At birth we have objectified a polypoid mass located in right minor labia and an anorectal malformation, vestibular fistula type. A colostomy, excision and biopsy of the mass were performed. Histological report informed of a fetal intermediate type rhabdomyoma. MRI showed a low anorectal malformation with fistula, right kidney slightly rotated and minimal pelvic ectasia, dysplastic coccyx, lipomatous lesion in conus medular, partial sacrum agenesis, probable uterus didelphys. Retrospectively, we reviewed our images and we didn’t observe the fetal anus; however we could identify a bicornuate uterus. At 23 days of life, it appeared a segmentary hemangioma in perineum and lumbosacral region orientating the case as a LUMBAR syndrome (Lower body infantile hemangioma, Urogenital anomalies and ulceration, Myelopathy, Bony deformities, Anorectal malformations, arterial anomalies and Renal anomalies).

Results

Although fetal perineal mass is rare, it can be detected by careful prenatal ultrasound. General counseling on diagnosis, treatment and prognosis of the mass should be given to parents. When detected, it is important to perform a precise evaluation of the fetal pelvic anatomy (uterus, urinary bladder, anus, sacrum, kidneys …) to rule out a more complex pelvic malformation and especially anorectal malformations. Prenatal sonographic diagnosis of anorectal atresia is often challenging and relies on indirect findings such as an abnormally dilated distal bowel segment which are not always present. The specific visualization of the fetal anus is a fairly reliable direct sign to rule out anorectal atresia. The anus is seen as a hypoechoic ring representing the perianal muscular complex surrounding an echoic central area central representing the anal canal mucosa. It should be correctly evaluated in cases of high risk of anorectal malformations (spinal anomalies, genitourinary malformations …). It is also useful to determine whether an anomaly is isolated or is part of a syndrome.

Conclusion

The prenatal diagnosis of a genital mass is rare. It seems reasonable to inform parents about the possible association with an anorectal malformation and be part of a syndrome. The prenatal diagnosis of anorectal atresia is feasible, but we have to think about it. The interest of our case relies on the exceptional association of a vulvar rhabdomyoma with a LUMBAR syndrome.