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A 20 days old patient with genital asymmetry

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A 20 days old patient reared as male was brought by his parents to Diabetes, Endocrine and Metabolism Pediatric clinic for atypical genitalia and bilateral undescended testis. On genital examination, the patient had underdeveloped right scrotal compartment, genital asymmetry, bifid scrotum, penoscrotal hypospadias and a phallus length of 3 cm. The left gonad could be felt at the medial end of the inguinal and it could be brought down to the scrotal sac and was of normal size and texture while the right gonad could not be felt along its course. The following investigations were ordered: Karyotyping, Basal hormones (17 (OH) progesterone, progesterone, DHEA, androstendione, testosterone, DHT, cortisol and ACTH) & Anti-Mullerian hormone and Abdominopelvic Ultrasonography. The result of the Karyotyping came back to show 45X0/46XY. Abdominal ultrasonography done revealed the presence of normal infantile uterus behind the urinary bladder, the vaginal canal was mildly dilated with fluid contents, a gonad mostly testis was seen on the left side measuring 12×7.2 mm and no gonad could be detected on the right side along its path of descent. The patient was prepared for laparoscopy at the department of Pediatric Surgery, Cairo University. During laparoscopy, specimens were obtained for histopathological examination and mullerian structures could be detected. The results of the histopathological examination revealed the presence of streak testis with male type ductal system which confirmed the diagnosis of mixed gonadal dysgenesis which is a variant of Turner Syndrome (Turner Syndrome with Y cell line)

Biography

Marise Abdou has joined Diabetes, Endocrine and Metabolism Pediatric Unit (DEMPU) in 2010 and has completed her MD from Cairo University. She is an active Member of DEMPU which was founded in 1980 by Prof. Dr, Isis Ghaly. She is actively involved in many research studies that are carried out in DEMPU. She has one publication in the field of endocrinology.

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