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Letter to Editor

Paediatrics Section

# Is Chromosomal Study Necessary for Girls with Inguinal Hernia?

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Dear Editor,

We read with great interest the recent case report by Konar S et al., [1]. They diagnosed an eight-year-old female presenting with bilateral inguinal hernia and a completely female phenotype as Complete Androgen Insensitivity Syndrome (CAIS) in that article. The authors suggested that screening for CAIS should be considered in all prepubertal girls presenting with inguinal swelling. We present the ultrasound findings of a newborn with a left inguinal mass from a paediatric endocrinology point of view.

A 10-day-old girl who had been born via normal spontaneous vaginal delivery at 39 weeks and with a birth weight of 3000 g was brought to our hospital by her family because of a palpable mass in the left groin. The case was evaluated at the paediatric endocrinology clinic after taking written consent from the family. The external genitalia were of female appearance but we thought that the gonad palpated in the left inguinal region could be the testis. Karyotype analysis was requested with a preliminary diagnosis of CAIS. Ultrasonography revealed that the uterus, 32x9x8 mm in size, was at the midline and the right ovary, 20x9x9 mm in size, was at its normal localisation. The left ovary was not at its normal localisation and a soft tissue area approximately 15x7.5x11 mm in size and consistent with ovary parenchyma was seen within the hernia sac in the left inguinal region. It contained 3-4 follicles with the largest 5 mm in size. The chromosome analysis result was 46,XX. The left ovary was reduced and hernia repair performed by the paediatric surgery department. We had requested karyotype analysis for the differential diagnosis before Ultra-Sonography (USG) was performed. However, we later decided there was no need for karyotype analysis and therefore prepared this letter accordingly.

CAIS is a rare condition with the estimated prevelance of 1:20000 and 1:99000 genetic male [2]. If one examines phenotypic females with inguinal hernias, the prevalence is noted to be 0.8% to 2.4% [2]. So, majority (> 97%) of phenotypic female cases presenting with inguinal hernia less likely to have CAIS. It has been suggested that all premenstrual girls with inguinal hernias should examined for CAIS [1]. If a testis is present in the hernia sac in a phenotypic female case presenting with inguinal hernia and there is no uterus on rectal examination, a preliminary diagnosis of CAIS can be considered. Another possibility is that the Mayer-Rokitansky-Kuster-Hauser (MRKH) patient, who will also have regression of the mullerian ducts and underdeveloped vagina. The physical examination will narrow the differential diagnosis and vaginal agenesis will most likely be due to the more common MRKH (46,XX) or CAIS (46,XY). We detected an ovary in the inguinal region in our case and the uterus was identified on USG. We therefore did not consider the MRKH but it should be considered in the differential diagnosis of CAIS in teenage cases presenting with primary amenorrhea. Current screening methods such as sonography or karyotype determination are time-consuming and costly and the purported incidence of CAIS has not been convincing enough to support their routine use [3]. Hurme T et al., diagnosed CAIS in only one case following karyotype analysis of 109 premenstrual females who had undergone inguinal hernia repair [4]. The first consideration when a gonad is palpated in the inguinal canal in paediatric endocrinology practice is therefore a testis, whether there is ambiguous genitalia or not, indicating a 46,XY sexual development disorder. However, there may be no need to perform chromosome analysis when a hernia sac containing the female reproductive organ(s) with a fully female external appearance. The uterus, cervix, fallopian tubes and proximal vagina will not develop in patients with a 46,XY chromosomal structure as there is a normal testis, testosterone production, conversion to dihydrotestosterone, and anti-Mullerian hormone. So, the vagina in CAIS is blind-ending, shortened and shows regression of the mullerian ducts. There may be no need for chromosome analysis and CAIS can be ruled out in general if there is a palpable gonad in the inguinal region in a patient with a female appearance and if ultrasonography reveals an ovary and uterus [5]. If no ovaries or fallopian tubes are found, consent for karyotyping should be sought [4].

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