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Reply to Letter to the Editor

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Dear Editor,

We thank Dr. Dolgin for his letter to the Editor addressing the incidence and diagnostic opportunities of complete androgen insensitivity syndrome (CAIS) in girls with inguinal hernia. We fully agree on the importance of timely recognition of CAIS, and we are pleased to elaborate on this topic.

CAIS is one of the disorders of sex development (DSD) and is caused by a mutation in the androgen receptor gene, located on the X chromosome. Because of the 46,XY genotype and the presence of testes, girls with CAIS do not have a uterus and proximal vagina. CAIS, however, is characterized by normal looking female genitalia and appearance, which make it hard to distinguish girls with CAIS from 46,XX girls. Consequently, CAIS often remains undiagnosed until late adolescence when girls may present with primary amenorrhea, or even later with infertility. A much earlier diagnosis enables timely genetic and parental counseling regarding several long-term issues e.g. infertility, hormone replacement therapy, potential sexual and psychological complications, the risk of malignant germ cell tumor, and appropriate scheduling of gonadectomy.

It has been reported that 90% of girls with CAIS undergo inguinal hernia repair during childhood [1]. Subsequently, inguinal hernia, or presence of a palpable inguinal or labial mass are noted as being the only harbingers of a potential diagnosis of CAIS in premenstrual girls. However, merely 57% of CAIS patients actually present with inguinal hernia of whom only a third has palpable gonads [2]. The incidence of CAIS in our study population (0.6%) might therefore be an underestimation, since we only identified patients who presented with inguinal hernia, and cases may have been missed when the testis or testes were not present in the hernia sac [3].

To increase the chance of detecting and diagnosing CAIS in premenstrual girls with inguinal hernia, we advise to always perform intraoperative exploration for gonadal tissue and Müllerian structures during hernia repair. If there is certainty about the presence of normal Müllerian structures, then there is no CAIS. However, if Müllerian structures are or seem to be absent, or their presence cannot be assessed during surgery, pelvic ultrasonography by a pediatric radiologist is indicated. Karyotyping finalizes the diagnostic process. We discourage routine preoperative ultrasonography, since the "number needed to diagnose" would be far too high and intraoperative examination of gonadal tissue is feasible (especially in the era of laparoscopy). We only recommend a preoperative ultrasound (followed by karyotyping in case of abnormal or inconclusive findings) in girls with bilateral hernia content during physical examination, since we showed that it is a major risk factor for CAIS [3].

Yours sincerely,

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