

Laparoscopy in inguinal hernia and complete androgen insensitivity syndrome in children. Whether and when to remove the gonads because of cancer?

Laparoskopia w operacji przepukliny pachwinowej u dzieci z zespołem niewrażliwości na androgeny. Czy i kiedy usuwać gonadę ze wskazań onkologicznych?

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Słowa kluczowe: laparoskopia, dzieci, przepuklina pachwinowa, zespół niewrażliwości na androgeny.

Abstract

Androgen insensitivity syndrome (CAIS) – also called Morris syndrome, formerly known as testicular feminisation syndrome – is a congenital disorder of sex development caused by various mutations in the gene encoding the androgen receptor. Androgen insensitivity syndrome occurs in a complete form (complete androgen insensitivity syndrome – CAIS) or a partial form (partial androgen insensitivity syndrome – PAIS). In approximately 1% (according to various researchers, 0.8–2.4%) of cases of inguinal hernia in girls during the time before menarche, CAIS is seen. The laparoscopic procedure for inguinal hernia repair allows a visual assessment of the gonads, and possibly the collection of specimens from the gonads. This is much simpler, compared to the conventional procedure.

Streszczenie

Zespół niewrażliwości na androgeny (zespół Morrisa, dawniej określany jako zespół feminizujących jąder) to wrodzone zaburzenie rozwoju płciowego spowodowane różnymi mutacjami w genie kodującym receptor wiążący androgeny. Zespół występuje w postaci kompletnej (*complete androgen insensitivity syndrome* – CAIS) albo częściowej (*partial androgen insensitivity syndrome* – PAIS). W ok. 1% (wg różnych autorów 0,8–2,4%) przypadków przepuklin pachwinowych u dziewczynek w okresie przed pierwszą miesiączką możemy mieć do czynienia z CAIS. Operacja laparoskopowa zaopatrzenia przepukliny pachwinowej umożliwia wizualną ocenę gonad i ewentualnie pobranie wycinków z gonad. Jest to o wiele prostsze niż w przypadku zabiegu metodą klasyczną.

Case report

A 2-year-old girl was admitted for elective surgery due to left-sided inguinal hernia. The surgery was performed by laparoscopic procedure using the percutaneous internal ring suture (PIRS) technique. Mini-laparotomy was performed through the umbilical port with optics of 3.5 mm or 5 mm and an angle of view of 30°. Wide, open orifices of both inguinal canals were visualised. Bilateral inguinal hernia was diagnosed. Gonads were observed bulging into both inguinal canals, their external appearance resembling testes more

than ovaries (Figures 1–2). Simultaneously, the uterus was not visualised (Figure 3). A second port, of 3.5 mm, was introduced at the right iliac fossa. Specimens were collected from the two gonads. The inguinal hernias were not repaired by the PIRS technique. Sutures were placed at the port sites. Vaginoscopy was performed, which revealed a very shallow vagina, without a visible cervix. No pathologies were observed in cystoscopy. The urethra, urinary bladder, and the urethral orifices were normal. Histopathology test result confirmed the presence of tissues characteristic of the texture of underdeveloped, infantile male gonad without



Figure 1. Left gonad (laparoscopic view)



Figure 2. Right gonad (laparoscopic view)

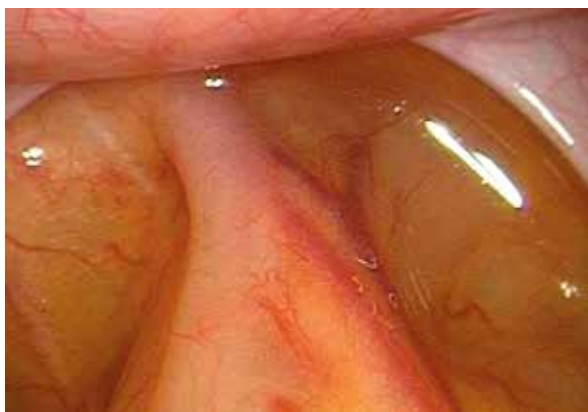


Figure 3. Laparoscopy – view of pelvis without a uterus

the features of germ cell proliferation. The child was subjected to genetic and endocrine diagnostics. In the genetic test, a male karyotype 46 xy was found, inconsistent with chronological gender (examination was performed after 72-hour culture of peripheral blood lymphocytes, 15 mitoses, type of staining GTG-500 bands). Examinations of hormone levels showed the following: oestradiol in blood plasma 0.00 ng/ml,

testosterone 0.00 ng/ml, FT4 0.84 ng/dl (normal value: 0.58–1.64 ng/dl), TSH 1.64 μ IU/ml (normal value: 0.34–5.6 μ IU/ml), FSH 3.91 mIU/ml (normal value: follicular phase 3.85–8.78 mIU/ml, periovulation period 4.54–22.51 mIU/ml, luteal phase 1.79–5.12 mIU/ml, after menopause 16.74–113.5 mIU/ml, lack of standards for children before menarche), and hLH 0.51 mIU/ml (normal value: follicular phase 2.12–10.89 mIU/ml, periovulation period 19.18–103.0 mIU/ml, luteal phase 1.2–12.86 mIU/ml, after menopause 10.87–58.64 mIU/ml, lack of standards for children before menarche). In ultrasonographic scan of the inguinal canals performed in a supine position the pathological structures were not visible. In a standing position a tissue area of testicular echostructure bulged into the upper part of the inguinal canals, on the right side, of size 14 \times 6 \times 8 mm and volume 0.4 ml; adjacent to it a layer of fluid up to 4.5 mm; and on the left side, a gonad of the dimensions 13 \times 6.5 \times 9 mm and volume 0.4 mm. As well as this, a bulging of the intestinal loop into the right inguinal canal was clearly observed (tension-free inguinal hernia). At present, the child remains under supervision and is being prepared for removal of the gonads after the development of the breasts; the parents decided about the management of the child in accordance with chronological gender. Three years have

elapsed since the diagnostic laparoscopy; paediatric surgeons and parents have not observed in the child the presence of inguinal hernias.

Discussion

The incidence of complete androgen insensitivity syndrome (CAIS) is 1 per 40,000–60,000 live births. It is diagnosed during the period of puberty due to primary amenorrhea, most frequently in girls with normally developing breasts, with sparse pubic and axillary hair [1–4]. Phenotypically, this is a woman; however, the vagina ends blindly and is shallow, and the uterus is absent [5–7]. The syndrome, first described in 1953 by Morris, was initially named ‘feminising testes syndrome’; subsequently this term was rejected as both offensive and incorrect. Testes in women with complete androgen insensitivity syndrome show a great deal of similarity to those observed in cryptorchidism, which suggests that anomalies in the testes are related with the abnormal position of the testes, rather than resistance to androgens. The testes may often be positioned within the inguinal canal, which may result in the formation of inguinal hernia. This has led to the conclusion that infants of female gender with bilateral inguinal hernias should be subjected to examinations for CAIS or the syndrome associated with the presence of the genotype XY [5–7]. Due to the fact that there is a low risk (3–5%) of gonadal carcinogenesis (the most frequent tumour is seminoma) up to the age of 25 [5, 8], the saving of the testes and their removal not before the period of puberty allows the natural development of the breasts, without the necessity for exogenous administration of oestrogens [5]. This is due to the transformation of androgens into oestrogens, and also allows the patient to make a conscious decision concerning the date and type of surgical procedure. Women with CAIS possess a feminine gender identity, and the majority of them are heterosexual. They have the potential to obtain normal sexual satisfaction. Nevertheless, in standardised questionnaires for the assessment of sexual satisfaction this was shown to be insufficient, mainly because of psychological maladjustment and length of the vagina (too shallow), which may be treated by vaginal dilatation. When this is ineffective, prolongation of the vagina may be obtained using a laparoscopic method [3, 5–7]. The majority of reports concern women during the period of puberty or later, when the major cause of reporting to a physician is primary absence of menstruation [1, 5, 9–12]. The occurrence of inguinal hernias, especially those that are bilateral, in infants of female gender and subsequently in girls, should draw the attention of the paediatric surgeon to the possibility of the occurrence of CAIS syndrome and its early diagnosis [2, 3, 13, 14]. This is especially important in the case of inguinal hernia repair by laparoscopic technique. This

technique allows direct observation of the gonads and possibly the collection of specimens from these gonads for histopathology tests [13–15]. It should be emphasised that in the case of performing the procedure by the conventional method, CAIS might not have been suspected because a unilateral inguinal hernia was dealt with, as confirmed by the presented study. Paediatric surgeons do not suspect CAIS syndrome in each case of bilateral hernias. Measuring the length of the vagina has even been proposed in order to make an early diagnosis of CAIS [2], in this way replacing a relatively complicated and costly imaging diagnostics when there is a suspicion of disorders in sexual differentiation. However, it is noteworthy that girls with CAIS usually do not have any external symptoms of the presence of male gonads. Although microtia was described in a patient with CAIS, these reports are casuistic [16]. At present, when an early diagnosis of CAIS is made, it is proposed that removal of the gonads is delayed until the time of normal development of the breasts and feminine phenotype [12]. Carcinomas related with undescended male gonads occur at an older age. These are primarily seminoma, dysgerminoma, or gonadoblastoma [6–8, 11]. Patients should undergo regular check-ups, as practised in patients with cryptorchidism. Early removal of the gonads results in the necessity for supplementation of oestrogens already in the early period of puberty in order to shape the feminine phenotype [8, 17].

Conclusions

The presented case of a girl with inguinal hernias (of which only one was observed by the parents, and the other was clinically silent), who had undergone PIRS procedure, allowed early suspicion of the occurrence of CAIS syndrome, as subsequently confirmed by additional tests. This enables further observation of the patient and the removal of the gonads by laparoscopic procedure, not before the normal shaping and development of the breasts.

Conflict of interest

The author declares no conflict of interest.

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