H-type anorectal malformations (embryology, anatomy, physiology, diagnosis).

Analysis of the literature.

Introduction. Patients who have a congenital rectourogenital connection and an external anal opening in a normal or ectopic position are considered to have an H-type malformation [1]. In a systematic review by Sharma and Gupta the reported incidence is 1-16% of all ARMs [2]. In Europe and America, this type of anorectal malformation (ARM) is considered rare. The European Anorectal Malformation Network (ARM-Net) found it in 0.5% of cases based on 10-year registration [3]. Lawal et al described 8 (0.7%) of 1170 females (1 day to 10 years) [4]. The H-type is 2,5-6 times more common in females [1,2]. In infants, it is diagnosed by the presence of meconium dots/streak on the perineum, in the vestibule of the vagina, or traces of meconium in the urine. Less frequently, the diagnosis was made due to frequent urinary tract infections before the age of 10 years [4]. H-type fistula should be suspected when there is a labial abscess and congenital fistula between labium/vulva and the anus [4, 5, 6,7]. Some authors describe only cases with the location of the fistula opening in the anal canal above or at the level of the dentate line [4, 5, 7, 8, 9,10]. For example, Li et al reported that "In all cases, the internal orifice opened above the dentate line" [10]. According to the Wingspread classification, H-type fistulas were divided into three groups according to their level. Low type double termination included cases in which the fistula was lying between the anal canal and the vestibule, and this was named as "perineal canal". In the intermediate type, communication was found between the rectum and the vestibule. High type of double termination consisted of a fistula between the rectum and the vagina [citation 11]. In males, the H-type is described not only with perineal ectopia of the anus, but also with urethral ectopia of different levels. For example, Rintala et al described three males who had a rectourethral H fistula and two males had a rectovesical H fistula [1]. When surgeons write about a normal anus or rectum in H-type ARM, they usually mean that it is open for bowel movements. However, in a study by Lawal et al, anal stenosis (caliber smaller than a size 9 Hegar dilator) was found in 3 of 8 patients [4]. I have not found any other studies of the anus and anal canal in this anomaly.

The aim of this study is to analyze the cases of H-type ARM published in the available literature from the point of view of the embryology of ARM development [12]. Below are 7 images of the perineum of patients with low H-type ARM. As an example of the norm, the image from the article by Amerstorfer et al [12] is taken **(Figure 1 a).**

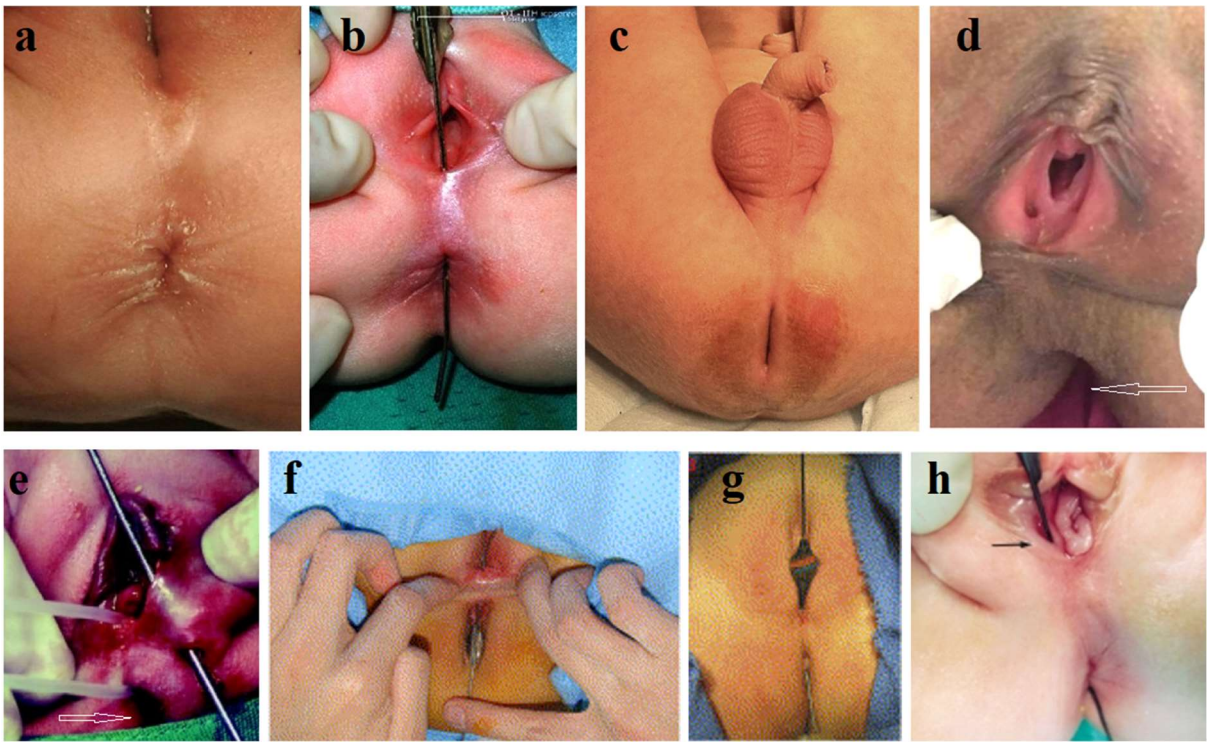

Figure 1. Perineal images of patients with H-type ARM. **(a)** Norma [12]. Skin folds on both sides of the closed anus due to contraction of the subcutaneous portion of the external anal sphincter (EAS). **(b)** Anovestibular fistula from the article by Sanal [13]. Funnel-shaped depression. **(c)** From the article by Slater et al [14]. Longitudinal cleft with no signs of EAS contraction. **(d)** From the article by Manjiri et al [5]. Perineal canal with anolabial fistula. Deep funnel instead of normal anus (arrow). **(e)** From the article by Yazlcl et al [15]. A light probe was pulled through the anovestibular fistula. The arrow shows the gaping anus. A metal sound was inserted through the left opening, and it was found that it had a subcutaneous continuity with a vulvar abscess. **(f)** From the article by Park [16]. Longitudinal gaping slit instead of anus. Catheter is pulled through anovestibular fistula. **(g)** From the article by Lawal et al [3]. Longitudinal slit without signs of contraction of the EAS. **(h)** From the article by Lee et al [17]. Oblique image shows a longitudinal, although not long, slit. A histological study in all 3 cases revealed squamous epithelium in the wall of the fistula, and the age of onset can confirm the "essentially congenital nature of the tract, evidence of inflammation being insignificant" [17].

Results

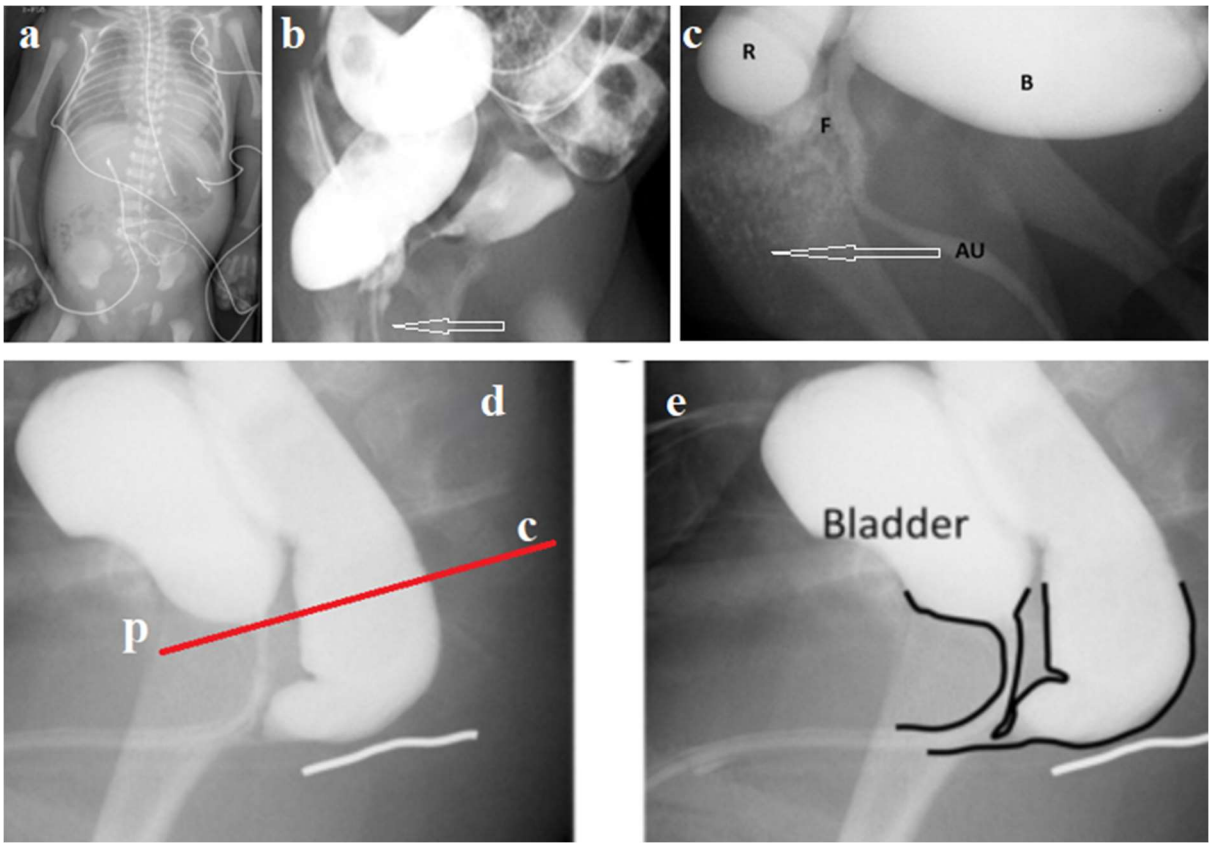

Figure 2. (a, b, c) The newborn boy had passed meconium in the first 24 h of life, but on exam was noted to have a slit-like anus (See Figure 1 c). Despite the anal stenosis (the anal opening was patent but admitted only a 9 mm Hegar dilator), the plain radiograph (a) shows that bowel movement was not difficult. An X-ray examination was performed, since particulate matter was also found in the urine. **(b)** During barium enema the ano-urethral fistula, described as a colourethral fistula, was found. The anal canal is in a contracted state (arrow). **(c)** Pre-operative voiding cystourethrogram demonstrating colourethral fistula. Contrast medium diluted with urine is seen in the widely open anal canal (arrow). During posterior sagittal anorectoplasty (PSARP), a "quite large and patulous" prostatic fistula was found. **(d-e)** From the article by Hosokawa et al [19], which shows the opening of the anal canal in ano-bulbar fistula in response to high pressure in the rectum. The fistula is not a canal, but an opening in the wall of the urethra. The distal wall of the anal canal is located a few millimeters from the anal fossa.

Analysis of the obtained results

All types of ARMs, excluding true cloaca, develop in the post-cloacal period, after the successful division of the cloaca with create the urethra, vagina, and anal canal. In the absence of associated urological pathology, the bladder, urethra, and vagina develop

normally. Initially, the IAS, within the endodermal part of the anal canal, descends through the perineal tissues towards the ectodermal rudiment. For an unknown (probably genetic) reason, the ectodermal primordium is not activated and does not form the distal part of the anal canal, resulting in the absence of an anus. By the time the IAS reaches the subcutaneous tissue opposite the anal dimple, it is located within the deep and superficial portions of the external anal sphincter, as well as in the loop of the puborectalis muscle and within the levator plates. The IAS, without encountering the ectodermal rudiment of the anal canal, moves forward and upward. In males, it can penetrate the perineum in front of the anal dimple, creating perineal ectopia, followed by urethral (bulbar, prostatic, and bladder-neck) ectopy. In females, it results in perineal, vestibular, and vaginal ectopy. The IAS becomes fixed at the site of penetration, away from the anal fossa to the fibrous ring, which is formed at the site of penetration of IAS through the skin and cutaneous tissue to the outside or through the wall of the organ (urethra or vagina). However, during defecation, because of contraction of the levator plates, its distal wall approaches the anal fossa and is located 2-4 mm from it, depending on age and the thickness of the skin and subcutaneous tissue. Therefore, when contracted, the length of the IAS is shorter, the higher its attachment. However, the length of the open anal canal corresponds to the age norm.

H-type ARM differs from the above-described situation by: (1) The damage to anus with frequent presence of stenosis; (2) a slit-like or funnel anus without skin folds on both sides indicate the absence of the subcutaneous part of the EAS. (c) The combination with anolabial fistula and labial abscess also indicates an unusual pathology of exogenous origin, since not all types of ARMs never shift from the central sagittal plane. (d) The presence of a functioning anal canal with an opening (fistula) on the anterior wall of the anal canal at different distances from the edge of the anus (from the dentate line in perineal ectopy to the upper third of the anal canal in vaginal ectopy and in ectopy in bladder-neck). (e) Significant predominance of females.

The embryology of this type of ARM can be presented as follows. Unusual damage to the exodermal rudiment of the anal canal leads to a slower formation of the anal canal to meet the endodermal rudiment. By this moment, the endodermal rudiment, not having met the exodermal one in time, began to shift in search of an exit outward or into the cavity of some organ **(Figure 3).** When the damaged exogenous rudiment joins the endogenous one at the level of the dentate line, the distal third of the anal canal is always pathological, although passable for feces.

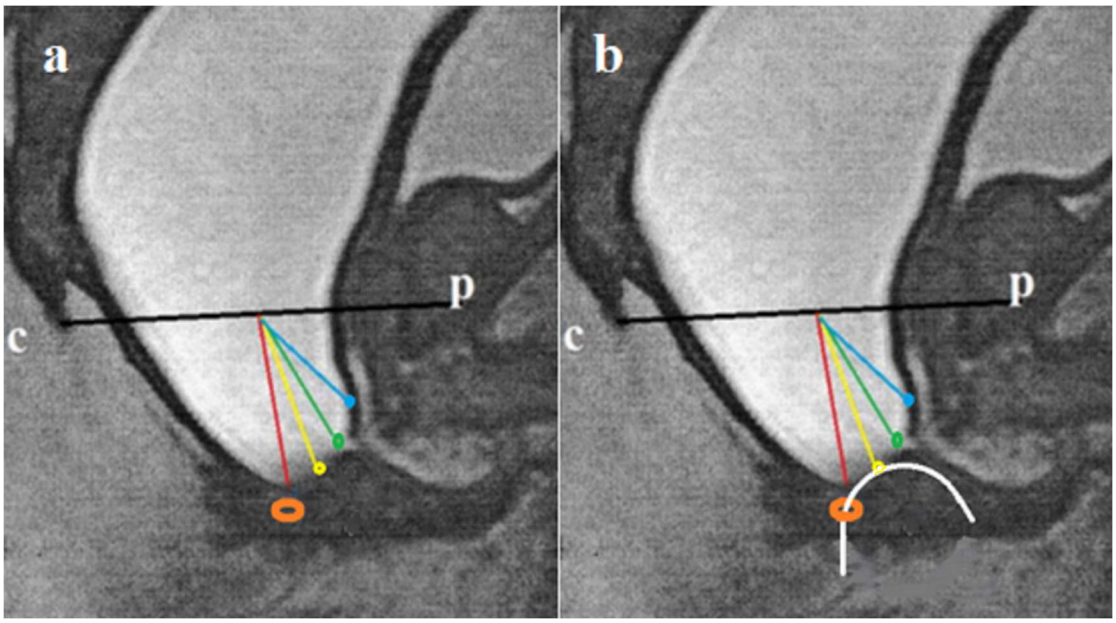

Figure 3. (a) For the diagram of possible variants of H-type ARM in males, a lateral anorectal CT with high-pressure contrast injection from a colostomy was used. High rectal pressure resulted in a wide opening of the anal canal below the pubococcygeal line. An anobulbar urethral fistula is visible as an opening in the anterior wall of the anal canal near the green dot. The distal wall of the anal canal is 2-4 mm from the anal verge, shown as an oval. The site in case of prostatic ectopy is shown in green, and perineal ectopy is shown in yellow. (b) Schematic diagram of ano-perineal ectopy. The catheter (white) inserted from the ectopic fistula (yellow) through the anus. At rest, when the anal canal is closed, the distance between the openings is smaller than in the diagram when the anal canal is wide open.

Anatomy of the so-called H-type ARM. Analysis of the literature shows that all types of ARMs without imperforated anus are ectopia of the anus on the anterior wall of the anal canal. The length of the fistula is equal to the thickness of the wall of the organ through which the IAS penetrates. The idea that the fistula is an extended channel with two openings is erroneous, which is why surgeons destroy the functioning anal canal without scientific justification [1,4, 13]. Based on anatomy, simple suturing of the fistula opening from the anal approach would be the most justified method. However, the mucous membrane in the fistula wall, as in the anal canal, is represented by squamous epithelium. The circumference of the fistula, clamped in the suture, cannot grow together and seal the fistula. Because of this, fistula relapse often occurs. Park utilized the endorectal mucosal advancement flap technique on three patients, and all had favorable outcomes [16]. Other surgeons successfully use anterior rectal wall pull-through to prevent relapses. “This technique involved sigmoidcolostomy, anterior rectal wall pull-

through, and followed by colostomy closure, which required 7–10 days of hospitalization” [5], Akhparov et al performed fistula extirpation and pull-through of the anterior wall [9]. The results were considered good if there was no recurrence of the fistula. However, the long-term results of the functions of fecal continence and defecation were not assessed. Experience in surgical treatment of ARM with visible fistulas suggests that to avoid recurrence, it is necessary to denature the cells of the mucous membrane of the fistula and the adjacent area of the anal canal near it and immerse them in a purse-string suture.

The name of the vice under discussion.

Currently, pediatric surgeons use different names: "H-type anorectal malformation", or as it is written in the Krickendeck classification - H fistula [1]. The term perineal canal was described by Stephen and Smith for an anorectal malformation in which a fistulous tract is noted between the anterior wall of a normally formed anal canal and the perineum [5]. Anovestibular fistula with normal anus [13]. Rectovestibular fistula with normal anus [10] Anorectal-vestibular fistula without an imperforate anus [7] H-type rectovestibular and rectovaginal fistulas [4]. H-type anovestibular fistula. [20]. Each of these names contains false ideas about the anatomy and physiology of this defect. Firstly, the use of the letter "H" suggests the presence of a long fistula with two openings, which is not true. Secondly, the statement about the normal anus is misleading, since in all the images we found, the anus had significant differences compared to the norm. In addition, the presence of stenosis in the anal canal has been repeatedly described. Thirdly, the use of the name "rectovestibular fistula" is based on the false assumption of the absence of the anal canal, which leads to the use of pull-through operations, which, regardless of access, destroy the functioning anal canal. The term perineal canal emphasizes its pathological essence but does not reflect its main features.

The name of the disease is a diagnosis that is determined because of a professional study, because the treatment tactics depend on it. Below are diagnoses that correspond to the anatomy and physiology of defects.

ARM. (Perineal) ectopy of the anus without an imperforate anus. In each case, the place of penetration of the anus (perineal, vestibular, vaginal, urethral - bulbar, prostatic or vesical) is inserted into brackets. Such a clarification as anovestibular fistula is correct, but does not make sense, since all options are at the level of the anal canal.

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