**Focus**

# The spectrum of anorectal malformations: a congenital disease for the general surgeons

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To be born with an anorectal malformation (ARM) is a rare event, estimated on 1 every 3000 live births. Many progresses have been made since the first reports by Paulus Aegineta,[1] but unfortunately two aspects are still unknow, or little known, that is the causes of this complex malformations and the ability to predict all possible associated malformations.

As for the esophagus, also the final part of the intestine, and in particular the anus, is little visible during pregnancy and, certainly, not at an early stage. Therefore the diagnosis of any sort of anorectal malformations is still at birth with obvious psychological impact on the neo-parents. The percentage of associated anomalies can be as high as 50% of cases, affecting, with different combination, the urologic tract, vertebral spine and spinal cord, heart and gastrointestinal system.[2] More systems are involved, more complex is the surgical repair, and more compromised can be, ultimately, the quality of life in the long term. Therefore, this kind of patients, throughout their entire life, will likely need different specialists to take care, time by time, of different problems. Pediatric surgeons and stoma therapist initially and until completion of surgical steps, pediatric urologists, neurosurgeon, orthopedist, social worker and/or psychologist at time of school, and gynecologist, urologist, colorectal surgeon and, again, psychologist since adolescence on. Besides medical and paramedical figures, the associations of affected patients or associations of patients sharing common problems, such as stomas and incontinence, are nowadays present throughout the world with the aim to help each other by collecting and sharing similar experiences and by providing neutral advices on the best centers of care.

A patient born in the last four or five decades with an ARM, either isolated or associated to other malformations, is expected to be alive nowadays and to face some of the problems listed above. The transition from pediatric to adult care for these patients is still largely inadequate almost around the world. Colorectal surgeons, urologist and gynecologists may be not prepared to be involved in the care of these patients who, conversely, may be reluctant to address any pediatric specialists. It is time, as for other malformations, pediatric and adult specialists get together thru workshops, websites and practice to guarantee an adequate level of competence to these patients. Such patients, indeed, may present a wide spectrum of complications which are relatively common conditions for pediatric surgeons, but are rarely seen by adult providers and a multidisciplinary approach is highly recommended for these complex cases.

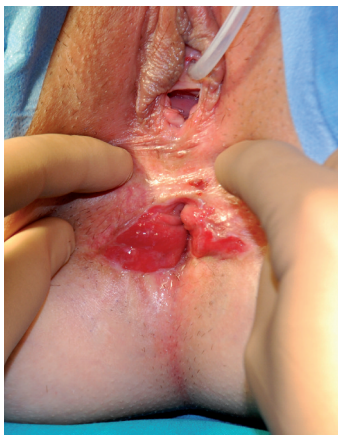

Figure 1. – Extensive ectropion of the anal mucosa.

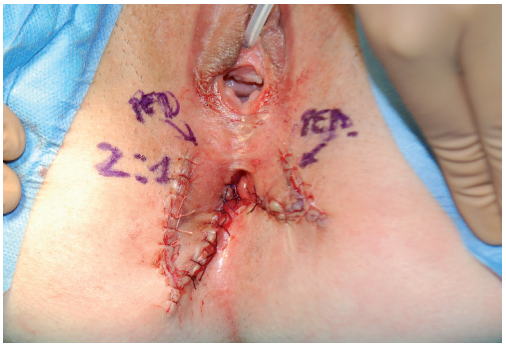


Figure 2. – Final aspect with mobilization of two V-Y flaps.

An example of such successful cooperation is herein reported. Sara was born 25 years ago with a recto-vestibular fistula, esophageal atresia, cardiac defects and asymmetric arms. At birth she underwent the esophageal repair and an "anorectoplasty", which consisted in the suture of the posterior wall of the rectum to the perineum, leaving the anterior wall ending into the vestibulum. At 3 years of age a formal anorectoplasty was performed under the protection of a colostomy and subsequently closed. She was followed by pediatric surgeons for a couple of years and then lost at follow up. At adulthood age she visited a colorectal surgeon because of fecal incontinence and an extensive ectropion of anal mucosa that limited her physical activity, attendance at work, and private life (Figure 1). The colorectal surgeon referred the patient to the pediatric surgeons of the center that treated her during childhood and together they examined her under sedation. At inspection the anus was patent, placed in the center of the sphincters, and an extensive portion of anal mucosa, inflamed and bleeding, extruded. Upon suggestion of the general surgeon, a plastic surgeon was consulted and a team composed by plastic and pediatric surgeons performed an anoplasty with resection of the prolapsed mucosa and mobilization of two V-Y flaps (Figure 2). The flaps healed very well and she could start using a mechanical system to empty the rectum, in order to control the fecal incontinence, and she is currently attending regular shifts at work.

The follow-up of anorectal malformations is life long and it may interest many specialists of adult care even if the malformations are treated during the first months of life by pediatric surgeons.

## REFERENCES

1. Paulus A: On the imperforate anus. In The Seven books, book VI, section LXXXI; translated by Adams F for the Sydenham Society, London 1944.
2. Alexander Matthias Holschneider and John M. Hutson. Anorectal Malformations in Children: Embriology, Diagnosis, Surgical Treatment, Follow-up. 2006 Ed.

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