Surgical treatment of abdominal wall weakness and lumbar hernias in Ehlers-Danlos syndrome – Case report

Oliver Scheufler [a,*], Julian Ramin Andresen [b], Reimer Andresen [c]

[a] Plastic and Aesthetic Surgery, AARE KLINIK, Bern, Switzerland
[b] Medical School, Sigmund Freud University, Vienna, Austria
[c] Institute of Diagnostic and Interventional Radiology/Neuroradiology, Westkuestenklinikum Heide, Academic Teaching Hospital of the Universities of Kiel, Luebeck and Hamburg, Heide, Germany

ARTICLE INFO

ABSTRACT

INTRODUCTION: Ehlers-Danlos syndrome (EDS) is a rare and diverse group of heritable connective tissue disorders. Gastrointestinal manifestations and abdominal pain are frequent in most subtypes of EDS. Conservative treatment is the standard of care.

PRESENTATION OF THE CASE: A 43-year-old female patient with genetically confirmed EDS classic subtype presented with diffuse gastrointestinal symptoms (bloating, belching and pain) that were controlled by the patient through inclined posture and external abdominal compression. A standard abdominoplasty with rectus muscle plication and mesh implantation lead immediately to complete relief of symptoms, which allowed the patient to assume an upright posture and resume all daily activities again. After 7 years, the patient was again seen with severe, persistent abdominal pain and inclined posture related to right lumbar herniations, as confirmed by MRI. However, there was no recurrence of the previous abdominal midline weakness and related gastrointestinal symptoms. Following lumbar hernia repair and mesh implantation, the patient was free of abdominal pain and resumed an upright posture again.

DISCUSSION: Although conservative treatment of EDS is primarily recommended and most surgeons are reluctant to operate on these patients except in life threatening situations, we present the successful surgical relief of disabling abdominal symptoms.

CONCLUSION: Regarding the variability and complexity of symptoms in different subtypes of EDS, a personalized multimodal treatment including surgical approaches should be considered and achieved a significant and long-lasting improvement in quality of life in our patient.

1. Introduction

Ehlers-Danlos syndrome (EDS), named after the dermatologists Edvard Ehlers and Henri-Alexandre Danlos, is a heterogenous group of hereditary connective tissue disorders characterized by varying degrees of skin hyperextensibility, joint hypermobility, and generalized skin fragility [1]. It may also affect vasculature, muscles, ligaments, tendons and internal organs. EDS results from defects in the synthesis of collagen with variable clinical presentations. The prevalence of EDS is reported to be one in 5.000–25.000 individuals worldwide but can be assumed to remain undiagnosed in many cases [2]. According to the Villefranche criteria, EDS is divided in 6 major subtypes: classic, hypermobility, vascular, kyphoscoliotic, arthrochalasia, dermatosparaxis [3]. The hypermobility type is most common, followed by the classic type; together these types account for 90% of cases [4]. Due to emerging scientific research and

discovery of new genes, a revision of this classification has been proposed [5]. In 2017, 13 subtypes of EDS were classified using specific diagnostic criteria [6].

Gastrointestinal symptoms related to EDS have been described in the past decade in multiple studies, including dysautonomia [7], hiatal hernia [8], rectal evacuation disorder [9], diverticulosis, gastritis [10], constipation, diarrhea, nausea, vomiting, abdominal pain, and reflux disease [11]. Nelson et al. found a prevalence of gastrointestinal symptoms in 58.9% of EDS classic, 57.5% of EDS hypermobility, 47.4% of EDS vascular and 43% of EDS others [12]. The most common gastrointestinal symptom in EDS overall was abdominal pain (43–82%) with 51.2% in EDS classic [12]. The prevalence of gastrointestinal manifestations of EDS appears to be higher than previously reported and may even lead to the diagnosis of EDS, such as in the rare case of visceral ptosis [13]. In a study by Zeitoun et al., 84% of patients suffered from functional bowel disorders and quality of life was significantly impacted by the presence of gastrointestinal symptoms as demonstrated by a poorer Gastroin-

* Corresponding author: AARE KLINIK, Florastrasse 13, 3005, Bern, Switzerland.
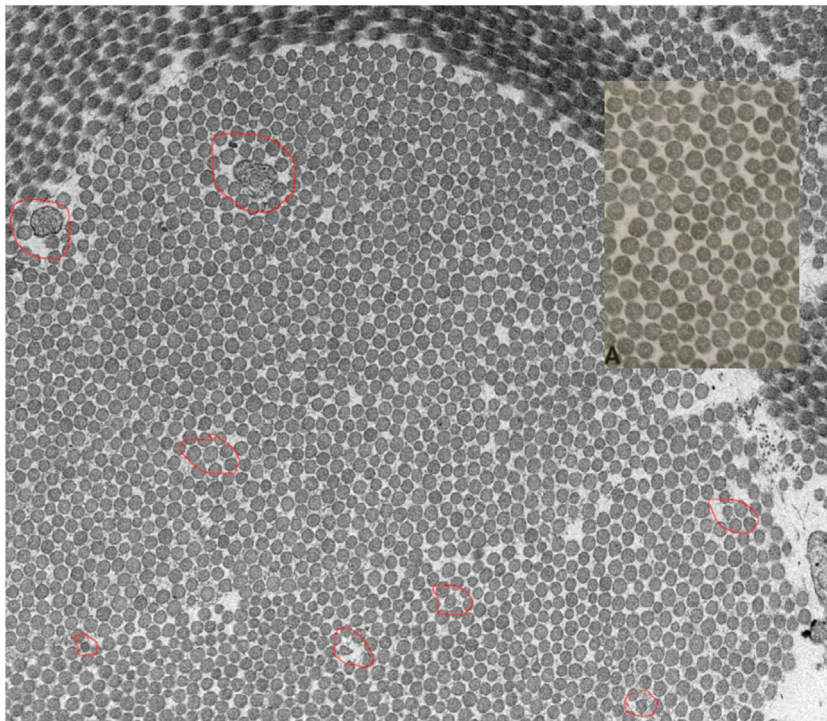E-mail address: oliver.scheufler@aareklinik.ch (O. Scheufler).

CASE REPORT – OPEN ACCESS

O. Scheufler et al.   International Journal of Surgery Case Reports 76 (2020) 14–18

Fig. 1. Electron microscopic image (magnification: 15.000x) with cross section of a collagen fibre bundle in the reticular dermis: multiple collagen fibrils show ultrastructural abnormalities, such as variable diameter, irregular shape and differing electron density.

testinal Quality of Life Index [14]. This work has been reported in line with the SCARE criteria [15].

2. Presentation of case

A 43-year-old female patient with a BMI of 23 and genetically confirmed EDS presented in 2012 with diffuse gastrointestinal symptoms, specifically bloating and uncontrollable belching in upright posture. She was able to alleviate these symptoms by assuming an inclined posture and external abdominal compression. The diagnosis of EDS classical subtype had been previously confirmed by electron microscopy (Fig. 1), biochemical collagen analysis and whole exome gene analysis, revealing mutations in COL5A1 and COL5A2. She had also been treated before for recurrent visceral ptosis by various pexy procedures. In her youth, the patient recalled a faster fatigue as the first clinical sign of her disease, a symptom that she also observed in her 6-year-old daughter that was also diagnosed for EDS. Visceral organ ptosis presented first as right nephroptosis and was treated by open nephropexy in 2000. Hepatoptosis with congenital lack of the right triangular hepatic ligament was corrected by laparoscopic hepatopexy with vicryl mesh and Argon beam induction of perihepatitis in 2002. Splenoptose was addressed by a laparosopic splenopexy in 2005 followed by diagnostic laparoscopy and adhesiolysis in 2006 and again in 2007, the latter combined with mobilization of the left colic flexure.

In addition, the patient was diagnosed for a symptomatic rheuma-factor negative (ANA and HLA-B27 negative) symmetrical polyarthritis in 2004. She also suffered from a chronic lumbospondylogenic pain with facet joint syndrome and erosive osteochondrosis Modic type I-II L5/S1 but without spondylodiscitis or neurocompression. The patient also suffered from posture related tachycardia that was thought to be related to an activation of the autonomic nervous system by visceral organ ptosis and had been treated with a β-blocker for 6 years.

During the initial consultation, the patient demonstrated abdominal bloating with uncontrollable belching in the upright posture that could only be alleviated by inclined posture and external abdominal compression. Also, chronic lower back pain resulted from permanent inclination and a previous disc herniation L5/S1 with chronic neck pain and muscle tension being triggered by the reclined head posture and hyperextension of the neck. On clinical examination in the supine position, there were no signs of bloating and the medial borders of the rectus abdominis muscles were palpable in the midline during contraction but were displaced laterally on relaxation, indicative of a rectus diastasis. The patient was able to withhold bloating and belching by voluntary abdominal muscle contraction with closing of the rectus diastasis. Because continuous muscle contraction was not feasible, assuming an inclined posture was the only other means for the patient to control her gastrointestinal symptoms. In addition, she wore a corset and used a cane when walking longer distances in order to relieve her back.

Therefore, we aimed for a permanent tightening of the abdominal wall. Preoperative blood and coagulation test were normal and a standard abdominoplasty with umbilical transposition, extended two-layered plication of the rectus aponeurosis in the midline and onlay VIPRO-mesh implantation covering the oblique and rectus abdominis muscles from the xiphoid to symphysis was performed (Fig. 2).

The surgery resulted in immediate and complete relief of the gastrointestinal symptoms, thereby allowing the patient to resume an upright posture with significant improvement of lower back and neck pain. The patient was instructed to wear a supportive abdominal belt for 3 months and to start muscle building exercises after 2 months. At 6 months follow-up, the patient reported no bloating, belching, back or neck pain. She had resumed her previous sports activities (dancing, skiing, rowing) and reported a significant improvement in quality of life after years of discomfort, pain and anxiety. Also, the tachycardia ceased and the β-blocker was discontinued.
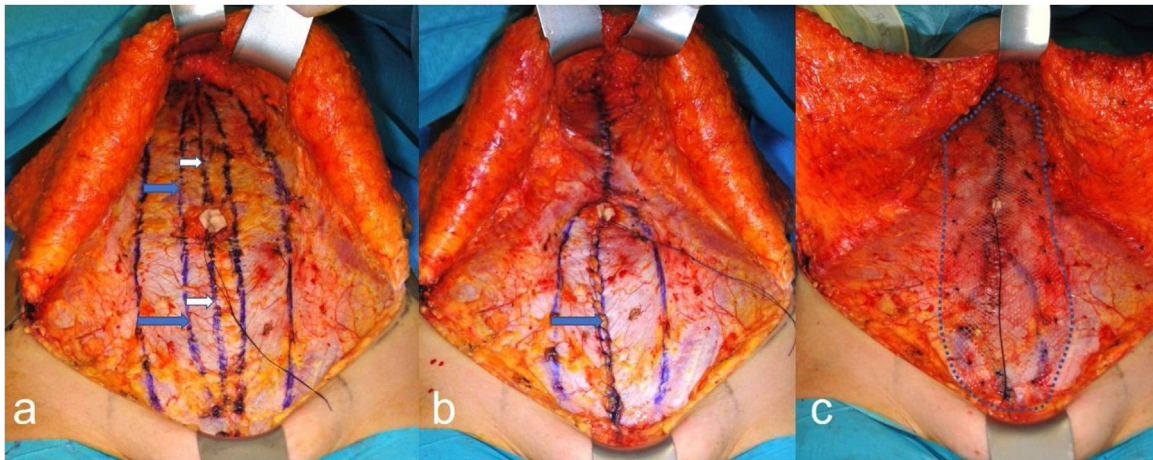
Fig. 2. Intraoperative photographs of abdominoplasty with two-layered plication of the rectus aponeurosis in the midline and onlay VIPRO-mesh implantation. In a), the midline, medial and lateral borders of the rectus muscle are marked with blue lines. White arrows indicate the medial and blue arrows the lateral aponeurotic plication. In b), the medial plication is shown in the lower abdomen, while the medial and lateral plication have already been completed in the upper abdomen. In c), the outer circumference of the VIPRO-mesh covering the rectus and external oblique abdominal muscles is marked by a blue dotted line.
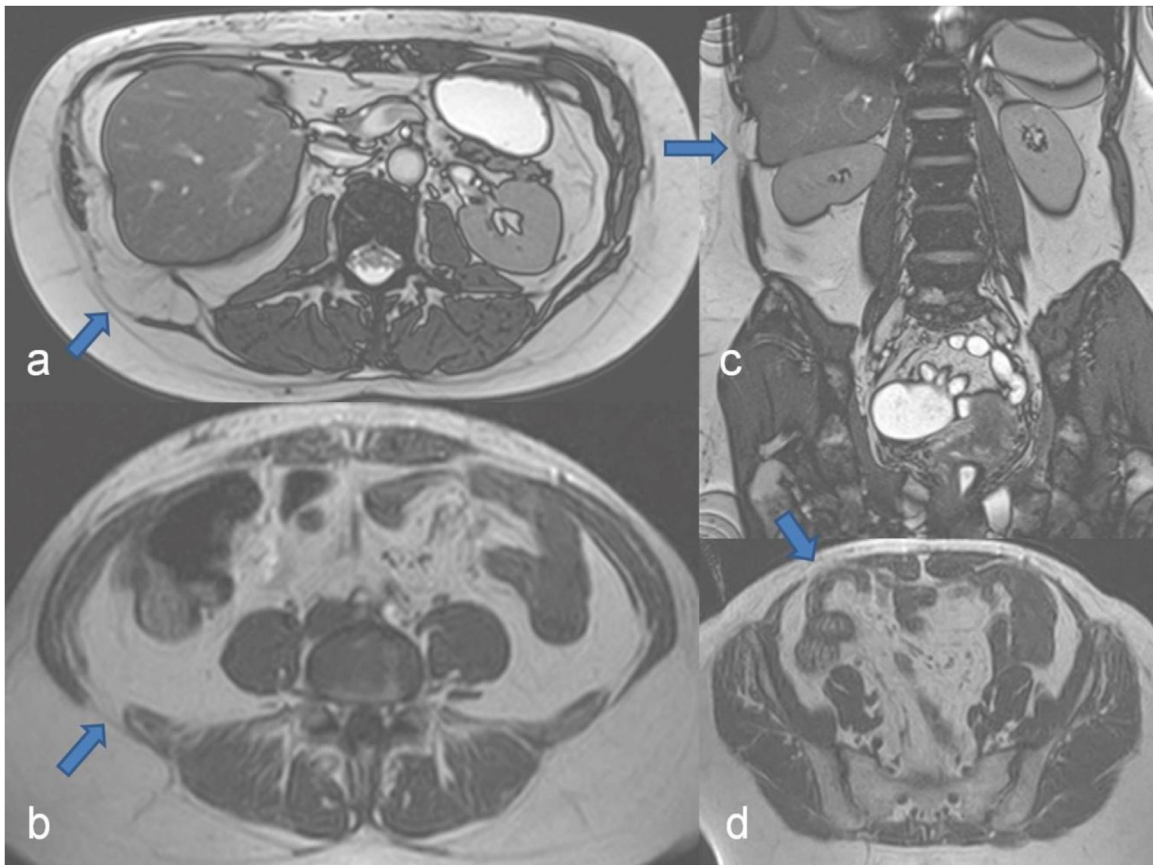

Fig. 3. Abdominal MRI with T2- and T1-weighted, no fat suppression images. In a) and c), the upper lumbar muscle defect with herniation of the visceral fat pad is marked with blue arrows (Grynfellt hernia). In b), the lower lumbar muscle defect is marked with blue arrow (Petit hernia). In d), a stable midline repair following rectus abdominis muscle plication and mesh implantation is visible. The blue arrow marks a defect at the lateral border of the rectus abdominis muscle as an incidental finding with no clinical symptoms.

In 2014, the patient underwent a diagnostic laparoscopy and left ovarian cystectomy with appendectomy. Two years later, she was again treated for a debilitating abdominal pain syndrome with pronounced slow-transit obstipation nonresponsive to laxatives and megacolon formation. The implantation of Tined Lead electrodes S3 for sacral neuromodulation in 2016 was not effective and was followed by total colectomy for pain relief.

The patient presented again in 2019 after a two-year period of persistent and aggravating right lumbar pain that was treated with morphine. Elevation of the right arm was painful and limited to below shoulder level. There was no abdominal, back or neck pain and no recurrence of previous gastrointestinal symptoms. However, the lumbar pain was debilitating and could be ameliorated only by wearing an abdominal compression belt and adopting an
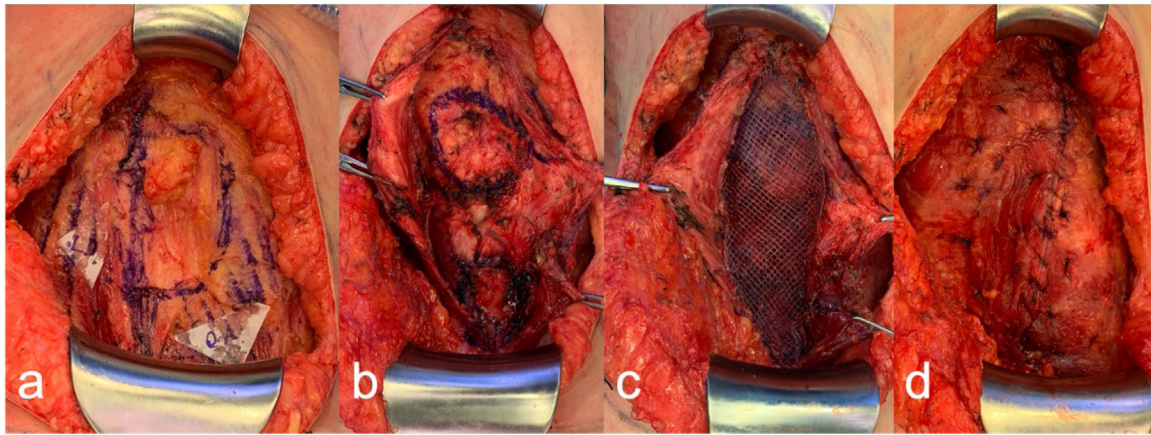
CASE REPORT – OPEN ACCESS

O. Scheufler et al.   International Journal of Surgery Case Reports 76 (2020) 14–18

Fig. 4. Intraoperative photographs of lumbar hernia repair. In a), the lumbar muscle defect between the latissimus dorsi muscle (LD) and external oblique abdominis muscle (OA) is marked. In b), the abdominal muscles are deflected with clamps and the upper and lower lumbar defects (marked in blue) are exposed. In c), the defects are covered with VIPRO-mesh and in d), the muscles are sutured over the mesh in a double layer.
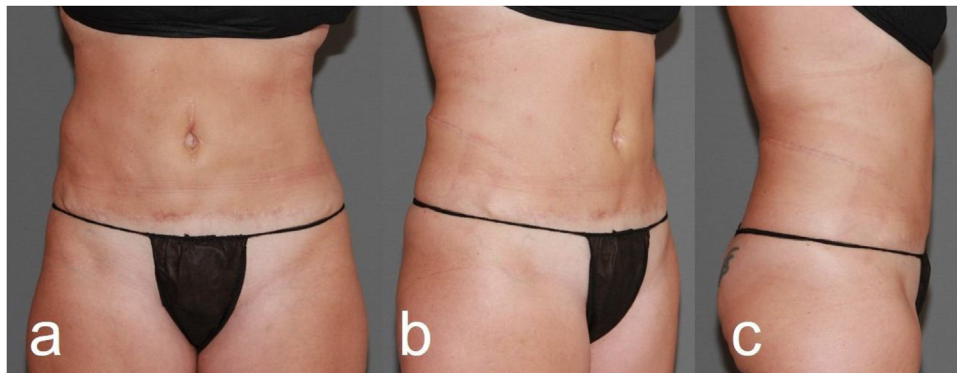

Fig. 5. Postoperative photographs 8 years after the abdominoplasty with rectus muscle plication and onlay VIPRO-mesh implantation and 1 year after lumbar hernia repair. The frontal (a), oblique (b) and lateral (c) views demonstrate a stable, long-lasting abdominal wall repair and inconspicuous scar formation on the lower abdomen and right flank.

inclined posture. A walking frame was used to cover longer distances.

Physical examination revealed a right lumbar swelling at the lumbotomy scar with moderate pain on palpation and pressure. Abdominal MRI in the standing and supine position visualized an interruption of the internal abdominal wall fascia between the serratus inferior posterior muscle and oblique internal abdominal muscle (Grynfellt hernia) with herniation of a fat lobe (6 × 3 cm cross-section) originating from the retroperitoneal space. There was also an interruption of the fascia between the atrophic latissimus dorsi muscle and the obliquus externus abdominis muscle (Petit hernia) with a gap of several centimeters (Fig. 3). These findings supported the clinical symptoms.

Because the hernias were not detectable laparoscopically, open repair of the superior and inferior lumbar hernias with insertion of a mesh was performed (Fig. 4). On the day after surgery, the patient began to ambulate in upright posture without the need of a walking frame. Healing was uneventful and the patient reported complete and persistent relief of lumbar pain and recaptured full elevation of the right arm above shoulder level at 1 year follow-up (Fig. 5).

3. Discussion

Surgical management of patients with EDS is challenging because of the rare disease and diverse subtypes with specific pathophysiology. Because of increased perioperative morbidity and delayed wound healing, conservative treatment is advocated as first line treatment, while surgery is usually reserved for life threatening and emergency indications, such as spontaneous bowel perforation or rupture of major arteries [16]. Easy bleeding and bruising are, to a variable degree, present in all subtypes of EDS caused by fragility of the capillaries and perivascular connective tissues as well as primary thrombocyte dysfunction and coagulation abnormalities [17,18]. They are characteristic manifestations in 25% of non-vascular EDS subtypes and up to 70% of EDS vascular due to primary thrombocyte dysfunction and vascular and skin fragility [18]. However, as pointed out by Castori, preventive contraindication to surgery is not justified in all EDS subtypes [19]. Less invasive laparoscopic procedures have been employed to reduce the morbidity from tissue friability and poor wound healing in EDS [20]. EDS specific recommendations for anesthesia and perioperative management have been advocated to minimize perioperative complications [21].

We present a patient with EDS classic that over the course of 20 years underwent multiple surgical procedures, including laparoscopic visceropexy and colonic surgery as well as open abdominal wall reconstruction for muscle diastasis and lumbar herniation. Considering the high surgical morbidity in EDS patients in the literature [16–21], there were no major perioperative complications regarding bleeding, infection or wound healing. Due to the severe and debilitating gastrointestinal manifestations of EDS, the patient was highly motivated to undergo repetitive surgery with long-lasting relief of complex symptoms and significant improvement in quality of life.

CASE REPORT – OPEN ACCESS

O. Scheufler et al. International Journal of Surgery Case Reports 76 (2020) 14–18

4. Conclusion

Regarding the complexity of symptoms in our patient with EDS classic, a personalized multimodal treatment including different surgical approaches was performed and achieved a significant and long-lasting improvement in quality of life. Our experience suggests the safety and reliability of laparoscopic and open surgical procedures in EDS classic in cases with severe gastrointestinal symptoms that cannot be managed conservatively. Our case indicates that patients with certain subtypes of EDS benefit, as a last resort, from surgical treatment when performed in a stepwise progression of invasiveness in debilitating gastrointestinal manifestations.

Declaration of Competing Interest

All authors declare no conflict of interest in formulating this article.

Funding

This research did not receive any specific grant from funding agencies in the public, commercial, or non-for-profit sectors.

Ethical approval

We declare that our institution does not require ethical approval of clinical case reports and that the study conforms to the ethical regulations of the declaration of Helsinki 1975 (revised current version).

Consent

Written informed consent was obtained from the patient for publication of this case report and accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal on request.

Author contribution

O.S. and R.A. contributed in conceptualization, study concept and design, O.S., J.R.A. and R.A. contributed in writing the paper.

Registration of research studies

1. Name of the registry: None.
2. Unique identifying number or registration ID: n/a.
3. Hyperlink to your specific registration (must be publicly accessible and will be checked): n/a.

Guarantor

The guarantor of this work, Oliver Scheufler, accepts full responsibility for the study and the conduct of the study, had access to the data, and controlled the decision to publish.

Provenance and peer review

Not commissioned, externally peer reviewed.

References

[1] J.L. Yen, S.P. Lin, M.R. Chen, D.M. Niu, Clinical features of Ehlers-Danlos syndrome, J. Formos. Med. Assoc. 105 (2006) 475–480.
[2] A.J. Hakim, A. Sahota, Joint hypermobility and skin elasticity: the hereditary disorders of connective tissue, Clin. Dermatol. 24 (2006) 521–533.
[3] P. Beighton, A. De Paepe, B. Steinmann, P. Tsipouras, R.J. Wenstrup, Ehlers-Danlos syndromes: revised nosology, Villefranche, 1997. Ehlers-Danlos National Foundation (USA) and Ehlers-Danlos Support Group (UK), Am. J. Med. Genet. 77 (1998) 31–37.
[4] B. Steinmann, P.M. Royce, A. Superti-Furga, The Ehlers-Danlos syndromes, in: B. Steinmann, P.M. Royce (Eds.), Connective Tissue and Its Heritable Disorders, Wiley-Liss Inc., Wilmington, DE, 2002, pp. 431–523.
[5] A. De Paepe, F. Malfait, The Ehlers-Danlos syndrome, a disorder with many faces, Clin. Genet. 82 (2012) 1–11.
[6] F. Malfait, C. Francomano, P. Byers, J. Belmont, B. Berglund, J. Black, et al., The 2017 international classification of the Ehlers-Danlos syndromes, Am. J. Med. Genet. C Semin. Med. Genet. 175 (2017) 8–26.
[7] Y. Gazit, A.M. Nahir, R. Grahame, G. Jacob, Dysautonomia in the joint hypermobility syndrome, Am. J. Med. 115 (2003) 33–40.
[8] Z.S. Al-Rawi, K.Y. Al-Dubaikel, H. Al-Sikafi, Joint mobility in people with hiatus hernia, Rheumatology 43 (2004) 574–576.
[9] S.D. Mohammed, P.J. Lunniss, N. Zarate, A.D. Farmer, R. Grahame, Q. Aziz, S.M. Scott, Joint hypermobility and rectal evacuatory dysfunction: an etiological link in abnormal connective tissue? Neurogastroenterol. Motil. 22 (2010), 1085-e283.
[10] M. Castori, F. Camerota, C. Celletti, C. Danese, V. Santilli, V.M. Saraceni, P. Grammatico, Natural history and manifestations of the hypermobility type Ehlers-Danlos syndrome: a pilot study on 21 patients, Am. J. Med. Genet. A 152A (2010) 556–564.
[11] N. Zarate, A.D. Farmer, R. Grahame, S.D. Mohammed, C.H. Knowles, S.M. Scott, Q. Aziz, Unexplained gastrointestinal symptoms and joint hypermobility: is connective tissue the missing links? Neurogastroenterol. Motil. 22 (2010), 252-e278.
[12] A.D. Nelson, M.A. Mouchli, N. Valentin, D. Deyle, P. Pichurin, A. Acosta, M. Camilleri, Ehlers-Danlos syndrome and gastrointestinal manifestations: a 20-year experience at Mayo Clinic, Neurogastroenterol. Motil. 27 (2015) 1657–1666.
[13] E. Reinstein, M. Pimentel, M. Pariani, S. Nemec, T. Sokol, D.L. Rimoin, Visceroptosis of the bowel in the hypermobility type of Ehlers-Danlos syndrome: presentation of a rare manifestation and review of the literature, Eur. J. Med. Genet. 55 (2012) 548–551.
[14] J.D. Zeitoun, J.H. Lefevre, V. de Parades, C. Sejourne, I. Sobhani, B. Coffin, C. Hamonet, Functional digestive symptoms and quality of life in patients with Ehlers-Danlos syndromes: results of a national cohort study on 134 patients, PLoS One 8 (2013), e880321.
[15] R.A. Agha, M.R. Borrelli, R. Farwana, K. Koshy, A. Fowler, D.P. Orgill, For the SCARE Group, The SCARE 2018 statement: updating consensus Surgical CAse REport (SCARE) guidelines, Int. J. Surg. 60 (2018) 132–136.
[16] J. Burcharth, J. Rosenberg, Gastrointestinal surgery and related complications in patients with Ehlers-Danlos syndrome: a systematic review, Dig. Surg. 29 (2012) 349–357.
[17] A. de Paepe, F. Malfait, Bleeding and bruising in patients with Ehlers-Danlos syndrome and other collagen vascular disorders, Br. J. Haematol. 127 (2004) 491–500.
[18] A. Anstey, K. Mayne, M. Winter, J. van de Pette, F.M. Pope, Platelet and coagulation studies in Ehlers-Danlos syndrome, Br. J. Dermatol. 125 (1991) 155–163.
[19] M. Castori, Surgical recommendations in Ehlers-Danlos syndrome(s) need patient classification: the example of Ehlers-Danlos syndrome hypermobility type (a.k.a. Joint hypermobility syndrome), Dig. Surg. 29 (2012) 453–455.
[20] G. Duthie, M. Singh, I. Jester, Laparoscopic management of colonic complications in Ehlers-Danlos syndrome type IV, J. Pediatr. Surg. 47 (2012) e1–e3.
[21] T. Wiesmann, M. Castori, F. Malfait, H. Wulf, Recommendations for anesthesia and perioperative management in patients with Ehlers-Danlos syndrome(s), Orphanet J. Rare Dis. 9 (2014) 109.