DOI: https://doi.org/10.17816/PED12285-89

儿童主静脉发育不良严重程度的研究

A CLINICAL CASE OF EXTREMELY SEVERE MAJOR VENES DISPLASIA IN A CHILD

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For citation: Azarov MV, Kupatadze DD, Nabokov VV, Makhin YuYu, Kolbaia LM, Dyug IV. A clinical case of extremely severe major venes displasia in a child. *Pediatrician (St. Petersburg).* 2021;12(2):85-89. https://doi.org/10.17816/PED12285-89

Received: 04.02.2021 Revised: 23.03.2021 Accepted: 23.04.2021

主静脉发育不良（DMV）是由描述这种病理的作者所知道的，即Klippel-Trenaunay综合征。在作者的经典描述中，Klippel-Trenaunay综合征的临床表现以三联征为特征：血管斑，非典型静脉曲张，软组织和骨骼肥大，患肢体积和长度增加。需要强调的是，这些症状的严重程度主要取决于病变的类型（胚胎或胎儿）和病变的严重程度。Klippel-Trenaunay综合征几乎总是散发性的，这意味着它在没有家族疾病史的人中发展。研究表明，这种情况是基因突变的结果，不是遗传的。这些被称为体细胞突变的基因变化，在出生前的早期发育阶段，在一个细胞中随机发生。Klippel-Trenaunay综合征可由*PIK3CA*基因突变引起。本文报道临床观察一一周岁儿童的病程，主静脉发育不良，症状极为严重。在目前的临床观察中，在以慢性疾病为主的背景下，有必要注意该患者的治疗难点。这些病人的治疗应在多学科医院进行，包括血管外科专家、整形外科医生和重症监护医生。以所述病例为例，说明了诊断策略和手术治疗。对大静脉发育不良患儿进行及时的病理手术和保守治疗，可以提高患儿的生活质量和社会适应能力。

关键词：血管畸形；Klippel-Trenaunay综合征；静脉；血管学；静脉造影术。

Dysplasia of the great veins (DMV) is known by the names of the authors who described this pathology as Klippel–Trenone syndrome. The clinical picture of the Klippel–Trenone syndrome in the classic description of the authors is characterized by a triad of symptoms: vascular spots, atypical varicose veins, hypertrophy of soft tissues and bones with an increase in the volume and length of the affected limb. It should be emphasized that the severity of these symptoms depends, first of all, on the type of lesion (embryonic or fetal) and the severity of the lesion. Klippel–Trenone syndrome is almost always sporadic, meaning that it develops in people with no family history of the disorder. Research shows that this condition is due to gene mutations that are not inherited. These genetic changes, called somatic mutations, occur randomly in a single cell during the early stages of development before birth. Klippel–Trenone syndrome can be caused by mutations in the *PIK3CA* gene. This article presents a clinical observation – the course of the disease of a 1-year-old child, with an extremely severe form of dysplasia of the great veins. In the presented clinical observation, attention is drawn to the difficulties of treating this patient against the background of the underlying chronic disease. The treatment of these patients should be carried out on the basis of a multidisciplinary hospital, which includes specialists in vascular surgery, an orthopedist and an intensive care physician. On the example of the described case, diagnostic tactics and surgical treatment are demonstrated. It is obvious that timely surgical and conservative treatment of pathology in children with dysplasia of the great veins improves the quality of life and social adaptation of children.

Keywords: vascular malformations; the Klippel–Trenone syndrome; veins; angiology; phlebography.

绪论

主静脉发育不良（DMV）是由描述这种病理的作者所知道的，即Klippel-Trenaunay综合征[2,7,8]。在作者的经典描述中，Klippel-Trenaunay综合征的临床表现以三联征为特征：血管斑，非典型静脉曲张，软组织和骨骼肥大，患肢体积和长度增加。Klippel-Trenaunay综合征几乎总是散发性的，这意味着它在没有家族疾病史的人中发展。这种缺陷，以及畸形和毛细血管畸形综合征是指由*PIK3CA*基因突变引起的过度生长综合征[1,10,11,20,22]。这种情况被称为与*PIK3CA*相关的过度生长现象（PROS）。对于专门从事这个问题的外科医生来说，全型Klippel-Trenaunay综合征和Weber综合征大瘘管病例中静脉和动静脉发育不良的鉴别诊断相对简单，然而，这些临床表现不明确的疾病的鉴别是非常困难的[3,4]。在描述Klippel-Trenaunay综合征患者的检查方法时，应该强调的是，目前静脉造影和较少使用的磁共振造影（MRI）对诊断和确定手术类型至关重要。磁共振检查和静脉造影结果100%与术中吻合[5,6,17,18,21]。

小儿主静脉发育不良的治疗一直是并仍是小儿外科手术中最困难的问题之一，这主要是由于该疾病的相对稀缺性、局限性和临床表现的广泛变异性[9,12,13,19]。重度主静脉发育不良患者的深静脉系统是由病理胚胎静脉池形成的（在几乎所有的病例中，下肢深静脉发育不全，血液流出通过单一的胚胎静脉）。很明显，病理过程中涉及的组织面积越大，外科医生就应该越谨慎[14]。治疗应是全面的，包括旨在防止慢性DIC综合征恶化的治疗[15,16]。对于病情极其严重的患者（畸形是慢性广泛性DIC综合征的特征）—肢体支持能力丧失，手术治疗是绝对需要的，在肢体丧失能力的情况下，不建议进行重建手术，同时确定具体的截肢指征。

临床观察

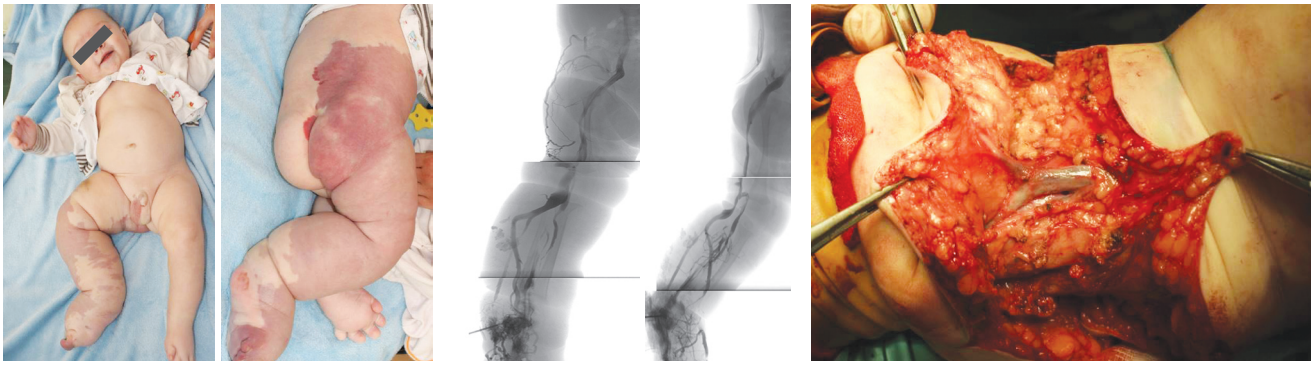
一名一岁男孩，一出生就一直生病，诊断为右下肢畸形肥大，左下肢和躯干有平滑的深红色血管斑点。大便中偶尔带一点血丝。大脚趾指甲板侧缘长入附近的软组织中，所以进行了切开排脓清疮治疗。患者1岁时在St.Petersburg State Pediatric Medical University显微外科观察。

检查发现：右下肢各节段明显增大，畸形肥大，有平滑的血管点通至腰椎和阴囊。摸到两个睾丸在阴囊里。左脚第一指间间隙较大，第二、三、四、五指间间隙增大。右手无名指明显增大。

实验室检查：临床血检、血液生化、常规尿检。凝血图：D-二聚体增加2.29 mg/L（正常值<0.5）。

左臀超声检查：伴静脉曲张的淋巴管瘤征象，9.6×2.74×7.69 cm。阴囊超声检查：双睾丸在阴囊内，右半囊水肿，有少量游离液体。右侧睾丸上方见一囊性回声。

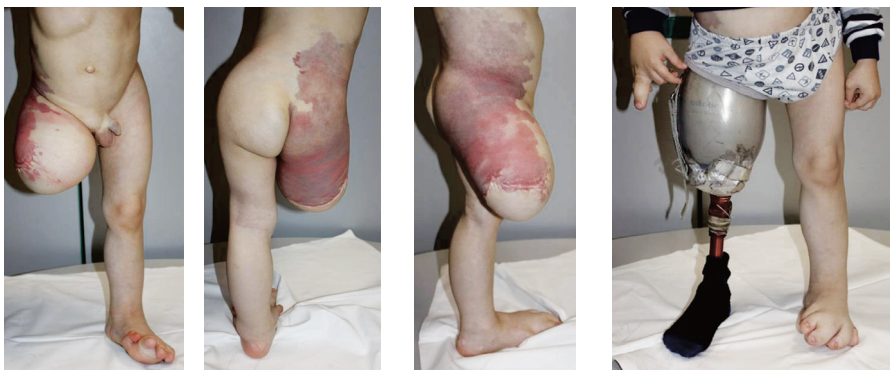
患者接受了右下肢骨内静脉造影（见图1）—下肢静脉和腘静脉段发育不全，动脉瘤样扩张，右侧髂股静脉段无明显对比增强，血液流出经过胚胎静脉系统，沿着小腿后外侧表面和大腿（坐骨神经移位）。



a *b* *c*

图. 1. K.V.患者，一岁：诊断为主静脉发育不良，胚胎型，极度严重：*a*—外观；*b*—顺行静脉造影术；*c*—腘区皮下组织胎儿静脉（手术一个阶段）

Fig. 1. Patient K.V., 1 years old: dysplasia of the main veins, embryonic type, extremely severe: *a* – appearance, *b* – ascending phlebography, *c* – embryonic vein in the subcutaneous tissue of the popliteal region (operation stage)



a *b* *c*

图. 2. K.V.患者， 两岁：诊断为主静脉发育不良，胚胎型，极度严重：*a*, *b*—手术后外观；*c*—带假体的外观

Fig. 2. Patient K.V., 2 years old: dysplasia of the main veins, embryonic type, extremely severe grade: *a*, *b* – appearance after surgery; *c* – appearance with a prosthesis

由于静脉血的流出是由位于皮下脂肪的血管进行的，因此没有进行血管外科矫正。在DIC综合征表现的背景下，深静脉的缺失，肌肉纤维化的进展和相关的关节挛缩，肢体功能的明显侵犯被认为是截肢的指征。经过与家长讨论，决定进行截肢手术。

两岁时，患者接受了右下肢髋关节水平截肢手术。右大腿切开了3个皮肤、皮下、腱膜的皮瓣（一个大的在健康皮肤的内侧表面，两个小的在外侧和后表面有血管斑点）。皮下组织严重纤维化。沿着后外侧表面移动一条直径达1.0 cm的大胚胎静脉，其对进行了结扎和穿过。由于血管纤维化，在技术上有困难，在大腿中部三分之一的股神经血管束被分离出来。神经周给药之后，进行了股神经穿过。股静脉缺失，在其凸出处有瘢痕，动脉无变化。血管被包扎、缝合和交叉。对坐骨神经进行了分离，神经周注射纳洛平溶液，神经穿过。股神经伴有直径达1.0 cm的大静脉（结扎，穿过），纤维改变的肌肉穿过，股骨被锯开，边缘被加工。采用电凝、组织缝合彻底止血。切除皮瓣用于闭合大腿末端缺损（大部分使用健康皮肤内侧皮瓣）。术后成功，创面基本愈合（见图2）。

术后早期观察到低蛋白血症（总蛋白44.3 g/L），在这方面进行了输血新鲜冷冻血浆，注射白蛋白。术后患儿在重症监护病房接受抗菌（头孢曲松）、止血（Tranexam、Dicynone）、输液、对症治疗，铁制剂，局部敷料。在术后期间，母亲注意到孩子的一般情况有了显著的改善。

结论

主静脉发育不良的外科治疗类型的选择应考虑疾病的严重程度。如果主静脉发育不良的程度非常严重，建议放弃进行重建手术，而接受精心调整的截肢手术。

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