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LETTER TO EDITOR

Klippel-Trénaunay-Syndrome in Childhood

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INTRODUCTION

Klippel and Trenaunay described in 1900 a rare congenital disorder of mesodermal development with dermal capillary malformations (most often a port wine stain over the affected limb, varicose dilatations and partial gigantism at one extremity [1]. Parkes and Weber described 1907 the same syndrome with further arteriovenous fistulas [2]. Klippel Trenaunay Syndrome (KTS) appears sporadically. The syndrome can appear at the upper and lower extremity, whereas the lower extremity is more often involved. An involvement of the breast and the abdomen was described [3,4]. Arteriovenous or capillary malformations will be found in more than 90% of all cases. Varicose malformations of the veins are found in 70% and partial gigantism in 60% of all cases [5]. The etiology is not exactly known. An autosomal recessive transmittance will be postulated, because KTS appears in female and male in the same proportion and appears sporadically [5-7]. Other authors suppose a genetic mutation. Several genetic mutations have been described in case series, but none of them have been proved to have any association with the disease. Genetic translocation at (8;14) (q22.3;q13), de novo supernumery ring chromosome 18, terminal deletion 2q37.3 and 5:11 balanced translocation have been described [8-10]. The incidence of KTS is two to five shells per 100.000 live births [11]. KTS was found in more than in one family member [9,11]. Different authors supposed an intrauterine injury of the intermediolateral tractus ending in a dilatation of capillary vessels [12,13]. A mesodermal and ectodermal dysplasia was presumed [6,10]. At birth the later apearance of KTS is nearly completely visible. The only changes, which are found in childhood are the growth of the extremity, the development of nodulary skin conditions and the fact that varices and lymphedema can be obviously be more present. Most often arteriovenous or capillary malformations are present [10,14,15]. The intensity of the colour can change over the years. Histologically vascular malformations with dilatated capillaries are found. Nodulary skin conditions are present extremely rare [10]. Hemangioma are also often described. Varicose dilatations of deep and superficial venes are not generally present at birth but can appear later. Venae perforantes can also be involved [7,11,16,17]. Anomalies of the deep venous system are known like duplications, hypoplasia, aplasia and compression of venes by connective tissue or deformed vessels [16,18]. Most often the vena poplitea and vena

femoralis are involved [19]. The degree of venous hypertension is directly related to the amount of collaterals. Some children develop thrombophlebitis. Moreover, intraabdominal and intrapelvic organs can be involved [3,4]. The hypertrophy of one extremity is the symptom of the most variability in KTS. The exact date, when growing of the extremity starts, is not exactly known. In many cases, a secondary lymphedema resulting out of malformations of the lymphatic system are the cause for the hypertrophy [19]. Overlapping cases with Sturge-Weber-Syndrome, spinal arterial malformations and Kasabach-Merritt syndrome do exist [1,3,19]. Cellulitis can be the consequence, which has to be treated by antibiotics in serious cases. An exact clinical and radiological evaluation of the extremity is necessary to perceive bone length differences and to start therapy options. Phlebography and MRI are used to prove varicose venes, embolisations and arteriovenous fistula. MR projection arteriography will also be used to explore the extent of vessel malformation [20]. Nonoperative procedures include intermittant pneumatic compression and compression bandages. Indications are chronic venous insufficiency, lymphedema, recurrent cellulitis and recurrent bleeding out of capillary and venous malformations [4]. Good results are seen, when IPC is performed in early childhood and when compliance of the patients is good. The size of the extremity can be reduced. Varicose venes and arteriovenous fistulae can regress. Stringel showed that in 81% of his cases, symptoms improved after IPC [4]. Long-term results after IPC in KTS are still lacking. Before superficial venes will be excised, the deep venous system must be proved for function [5,14-16]. The most often performed procedures are the ligation and excision of venes and stripping of the venes. In a study of Lindauer in 18 patients with KTS, which were treated surgically, in 92% the operation showed no satisfactory results [8]. The cause for these bad results were hypertrophic scarring in the soft tissue. Excision of extending venous and lymphatic malformations is difficult and can induce a high intraoperative blood loss. The advantages of excision must be compared with factors like postoperative pain, recurrent infections and bleeding. Rarely a complete excision of extending malformations is possible. Laser Therapy (Neodym-YAG) can diminish the colour intensity of capillary malformations. Most often repeated laser treatment is necessary. Laser Therapy is not successful in each case. Percutaneous Sclerotherapy can be performed in special cases with symptomatic venous malformations [17]. Sotradecole, ethanolamine and ethylalcohole will be used. 20% of all cases with KTS have no deep venous system. In this cases sclerotherapy of superficial venes should not performed. Complications after sclerotherapy are necrosis of the skin, nerve injury, hemoglobinuria and allergic reactions with anaphylaxia [4]. Because of less soft-tissue trauma the success rate after sclerotherapy compared to excision or varicose stripping is higher. Complications of Klippel-Trenaunay syndrome can include cellulitis, swelling caused by lymphedema and internal bleeding from abnormal blood vessels, pulmonary embolism and complications during delivery of a pregnant women [2,6,16,18]. A case with portosystemic encephalopathy was found in literature [21]. The condition is also associated with fusion of certain fingers or toes like syndactyly or the presence of extra digits, called polydactyly. Prognosis is good, most of the patients were generally treated conservative.

REFERENCES

1. Purkait R, Samanta T, Sinhamahapatra T, et al. (2011) Overlap of sturge-weber syndrome and klippel-trenaunay syndrome. Indian Journal of Dermatology 56(6): 755-757.
2. Douma RA, Oduber CE, Gerdes VE, et al. (2012) Chronic pulmonary embolism in klippel-trenaunay syndrome. Journal of the American Academy of Dermatology 66(1): 71-77.

3. Rohany M, Shaibani A, Arafat O, et al. (2007) Spinal arteriovenous malformations associated with klippel-trenaunay-weber syndrome: A literature search and report of two cases. AJNR American Journal of Neuroradiology 28(3): 584-589.
4. Robertson DJ (1956) Congenital arteriovenous fistulae of the extremities. Annals of Royal College of Surgeons of England 18(2): 73-98.
5. Ceballos-Quintal JM, Pinto-Escalante D, Castillo-Zapata I (1996) A new case of klippel-trenaunay-weber (KTW) syndrome: Evidence of autosomal dominant inheritance. American Journal of Medical Genetics 63(3): 426-427.
6. Jacob AG, Driscoll DJ, Shaughnessy WJ, et al. (1998) Klippel-trenaunay syndrome: Spectrum and management. Mayo Clinic Proceedings 73(1): 28-36.
7. Cohen MM (2000) Klippel-trenaunay syndrome. American Journal of Medical Genetics 93(3): 171-175.
8. Chen D, Li L, Tu X, et al. (2013) Functional characterization of klippel-trenaunay syndrome gene AGGF1 identifies a novel angiogenic signaling pathway for specification of vein differentiation and angiogenesis during embryogenesis. Human Molecular Genetics 22(5): 963-976.
9. Wang Q, Timur AA, Szafranski P, et al. (2001) Identification and molecular characterization of de novo translocation t(8;14) (q22.3;q13) associated with a vascular and tissue overgrowth syndrome. Cytogenetics and Cell Genetics 95(3-4): 183-188.
10. Brouillard P, Vikkula M (2007) Genetic causes of vascular malformations. Human Molecular Genetics 16 Spec No. 2: R140-R149.
11. Berry SA, Peterson C, Mize W, et al. (1998) Klippel-trenaunay syndrome. American Journal of Medical Genetics 79(4): 319-326.
12. Phadke SR (2009) Klippel trenaunay syndrome. Huret JL (Eds.), Atlas of Genetics and Cytogenetics in Oncology and Haematology: 153-155.
13. Kashiwada T, Fukuhara S, Terai K, et al. (2015) ß-catenin-dependent transcription is central to Bmp-mediated formation of venous vessels. Development 142 (3): 497-509.
14. Servelle M (1985) Klippel and trenaunay's syndrome. 768 operated cases. Annals of Surgery 201(3): 365-373.
15. Güngor Gündogan T, Jacquemyn Y (2010) Klippel-trenaunay syndrome and pregnancy. Obstetrics and Gynecology International 2010: 706850.
16. Neubert AG, Golden MA, Rose NC (1995) Kasabach-merritt coagulopathy complicating klippel-trenaunay-weber syndrome in pregnancy. Obstetrics and Gynecology 85(5.2): 831-833
17. Cabrera J, Cabrera J, Garcia-Olmedo MA, et al. (2003) Treatment of venous malformations with sclerosant in microfoam form. JAMA Dermatology 139(11): 1409-1416.
18. Huiras EE, Barnes CJ, Eichenfield LF, et al. (2005) Pulmonary thromboembolism associated with klippel-trenaunay syndrome. Pediatrics 116(4): e596-600.
19. Kasabach HH, Merritt KK (1940) Capillary hemangioma with extensive purpura: A report of a case. The American Journal of Diseases of Children 59: 1063-1070.
20. Ziyeh S, Spreer J, Rossler J, et al. (2004) Parkes weber or klippel-trenaunay syndrome? Non-invasive diagnosis with MR projection angiography. European Radiology 14(11): 2025-2029.
21. Yazaki M, Kaneko K, Tojo K, et al. (2008) An unusual case of klippel-trénaunay-weber syndrome presenting with portosystemic encephalopathy. Internal Medicine 47(18): 1621-1625.