Klippel-Trenaunay syndrome – a promising role of sirolimus? Case report

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Introduction: Klippel-Trenaunay Syndrome (KTS) is rare syndrome characterized by a presence of capillary and venous malformation, limb overgrowth, with or without lymphatic anomalies[1]. KTS is related to mutations in the PIK3CA gene. Complications of KTS include clotting disorder, bleeding, lymphedema, soft tissue infection and pain. Treatment is individualized; it may include endovascular or surgical procedures and supportive care (management of coagulopathy, infection, pain). The use of mammalian target of rapamycin (mTOR) inhibitor sirolimus shows promising results in complex vascular malformations[2,3].

Case report: We present a 45 years old patient, currently living out of Croatia. Vascular malformation of the left arm was noticed a few months after his birth. During his childhood an angiography of the left arm lead to confirmation of KTS without further specific treatment. During the last 15 years he was undergoing regular hematology controls and treatment of consumptive coagulopathy as complication of KTS. In 2010 at age 35, he suffered a fracture on the left arm, but he was declared inoperable on basis of vascular malformation which extended the healing process. From 2018 further proliferation of vascular malformation was noticed on his left hemithorax. In 2020 he was presented to multidisciplinary vascular team, MR angiography was performed Unfortunately, due to extreme extension of vascular malformation with soft tissue hypertrophy and osseous deformation the patient is not suitable for any surgical or endovascular procedure. We suggested a treatment with sirolimus which is delayed due to patient's temporary relocation. Genetic panel testing covering PIK3CA gene is in progress.

Conclusion: By presenting this case, our objective is to increase the awareness of KTS, related complications and the role of sirolimus which can improve the prognosis of vascular anomalies. Additionally, we wish to emphasize the importance of multidisciplinary vs specialty focused approach in management patients with KTS.

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LITERATURE

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