

Klippel–Trenaunay syndrome

Kamal Kant, Richa Purohit

CASE REPORT

A 17-year-old male with history of repeated surgeries for cystic lesions in neck and lower limb since infancy, abnormal overgrowth of toes and limbs, anaemia requiring multiple transfusions. On examination, young male, anaemic short statured with lower limb disabilities (Figure 1). Abdominal examination revealed huge splenomegaly, no ascitis, no other organomegaly. Scrotum contained soft cystic swelling bilaterally (Figure 2) transilluminant and no impulse. Left lower leg was deformed with scars all over as result of previous surgeries, overgrowth of toes and black naevus on foot (Figure 3). Patient was evaluated biochemically and by imaging USG, MRI of abdomen. MRI images (Figures 4–7) confirmed the diagnosis of Klippel–Trenaunay syndrome. Patient underwent splenectomy and was in follow up for one year.

DISCUSSION

The syndrome is a genetically inherited condition affecting the development of blood vessels, soft tissues and bones. It affects 1 in 100,000 populations [1] all over the world. It is caused by genetic mutation most commonly affecting PIKCA gene which is responsible for development of tissues in the body resulting in overgrowth.

Kamal Kant¹, Richa Purohit²

Affiliation: ¹MS, FACS, FRCS (Glasgow, UK), Sr. Consultant, Surgery, Medipulse Hospital, Jodhpur, Rajasthan, India 342003, B 24, Shastri Nagar, Jodhpur, Rajasthan, India; ²MS, MRCS (UK), Consultant Surgery, Dr. S.N. Medical College, Jodhpur, Rajasthan, India.

Corresponding Author: Dr. Kamal Kant, B 24, Shastri Nagar, Jodhpur, Rajasthan, India 342003; Email: kkant54@gmail.com

Received: 31 March 2018

Accepted: 20 April 2018

Published: 07 May 2018



Figure 1: Hypertrophied toes and deformed limb.



Figure 2: Scars of surgery in leg and lymph varix in scrotum.



Figure 3: Black naevus on foot.

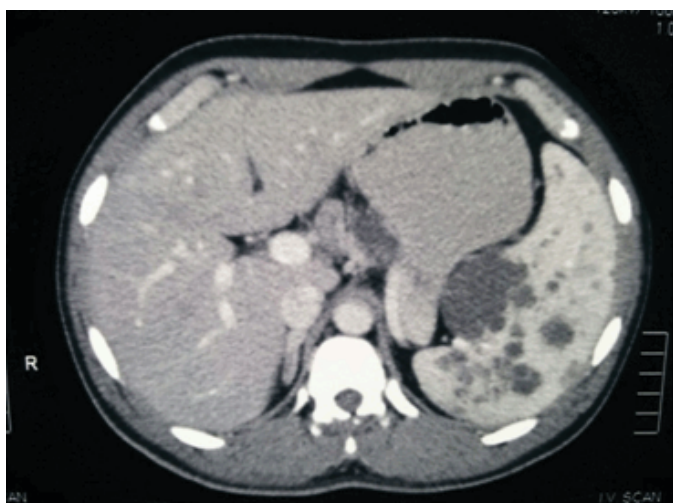


Figure 4: CT Image showing cystic lesions in spleen.

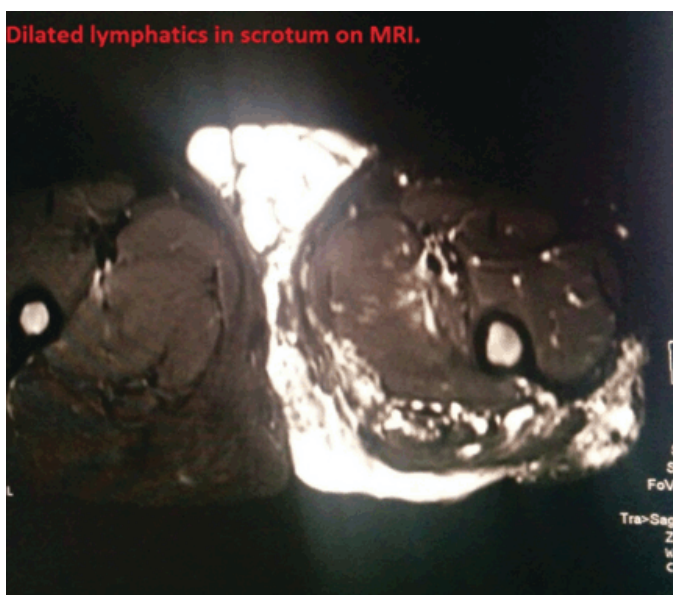


Figure 5: MRI of pelvis showing dilated lymphatics.



Figure 6: MRI of lower limb showing dilated lymphatics.

Common presenting features included:

A red birthmark called as Port wine stain. Abnormal overgrowth of soft tissues and bones. Venous malformation common in lower limbs. Lymphatic cysts all over viscera with lymphangiectasis as seen in the present case and anaemia of unknown cause, possibly splenomegaly. In the present case, all features of KT syndrome were present except the red port wine spot. Instead, a black naevus was present on foot as shown in the image.

CONCLUSION

This syndrome is a genetically inherited rare condition affecting malformation of soft tissue including blood vessels, bones, lymphatics in limbs and viscera. No definite treatment is available.



Figure 7: X-ray of leg showing deformed bones.

Article ID: 100027S05KK2018

doi: 10.5348/100027S05KK2018CL

Author Contributions

Kamal Kant – Substantial contributions to conception and design, Acquisition of data, Analysis and interpretation of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published

Richa Purohit – Substantial contributions to conception and design, Acquisition of data, Analysis and interpretation of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published

Guarantor of Submission

The corresponding author is the guarantor of submission.

Source of Support

None

Consent Statement

Written informed consent was obtained from the patient for publication of this clinical image.

Conflict of Interest

Author declares no conflict of interest.

Copyright

© 2018 Kamal Kant et al. This article is distributed under the terms of Creative Commons Attribution License which permits unrestricted use, distribution and reproduction in any medium provided the original author(s) and original publisher are properly credited. Please see the copyright policy on the journal website for more information.

REFERENCES

1. Reddy OJ, Gafoor JA. Rajanikanth M, Prasad PO. Klippel-Trenaunay syndrome with review of literature. J NTR Univ Health Sci 2015;4(2):120–3.

Keywords: Cystic swelling, Genetic mutation, Klippel–Trenaunay syndrome

How to cite this article

Kant K, Purohit R. Klippel–Trenaunay syndrome. Edorium J Surg 2018;5:100027S05KK2018.

Access full text article on
other devices



Access PDF of article on
other devices

