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.
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 lymphangiactasis 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.

REFERENCES


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

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Guarantor of Submission

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

Conflict of Interest

Author declares no conflict of interest.

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