

**TITLE OF THE ABSTRACT:** Clinical profile of cutaneous vascular anomalies and the associated overgrowth syndromes

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**Objectives:** The primary objective was to study the clinical features of cutaneous vascular anomalies and the overgrowth syndromes. The secondary objective was to describe extracutaneous features of vascular anomalies and the overgrowth syndromes.

**Methodology:** Patients with clinical features of cutaneous vascular anomalies were recruited in the cross sectional study between September 2014 to July 2016 after obtaining informed consent from patients and guardians in case of children. Imaging studies were done in majority patients wherever necessary to confirm the diagnosis and extent of involvement of vascular anomalies. Skin biopsies were done in few doubtful cases. Patients were classified according to the 2014 ISSVA classification of vascular anomalies. Data on clinical and radiological features were expressed in numbers and percentages. *P* value was calculated using Fisher's exact test wherever applicable.

**Results:** A total of 136 patients with cutaneous vascular anomalies were included in the study. There were a total of 60 males and 76 females (M:F-1:1.25). The hospital based prevalence of vascular anomalies was 0.89%. The mean age of patients presented with vascular anomalies was  $11.96 \pm 16.61$  yrs (range 1 month-73years). Vascular malformations (n=88, 65%) were more common as compared to vascular tumours (n=48, 35%). IH (n=42, 30.9%) was the most common vascular tumour followed by RICH (n=3, 2.2%), NICH (n=1, 0.74%), TA (n=1, 0.74%) and KHE (n=1, 0.74%). The only syndrome associated with IH in our study was PHACES syndrome found in 2 patients. The most common type of vascular malformation was capillary malformation (n=34, 25%) followed by LM (n=14, 10.3%), VM (n=10, 7.4%), mixed vascular malformations (n=7, 5.1%) and AVM (n=6, 4.4%). The rare overgrowth syndromes found in our study were PROS (n=8, 5.9%) including KTS (n=5, 3.7%) and CLOVES (n=2, 1.5%). The other rare syndromes associated with vascular malformations were PPV (n=5, 3.7%), SWS (n=4, 2.94%), HHT (n=1, 0.74%) and BRBB (n=1, 0.74%). The extracutaneous systems involved in our patients with vascular anomalies were skeletal system (21, 15.4%), CNS (9, 6.6%), eye (7, 5.15%) and abdomen (3, 2.2%). D-dimer levels were done in 17 patients out of which 9 patients had elevated levels. MRI and Doppler US were done in 27 patients each. Lateral marginal veins and phleboliths were found in 3 patients each

**Conclusion:** Vascular malformations were more commoner than vascular tumours. Female preponderance was noted in all subtypes of vascular tumours. IH was the most common cutaneous vascular tumour. Most of the IH involved head and neck region. PHACES syndrome was found in 2 patients with large facial IH, posterior fossa

structural and cerebrovascular abnormalities. The only case of KHE was associated with KMP. Male preponderance was noted in all subtypes of vascular malformations except venous malformations where females were predominant. Capillary malformation was the most prevalent type of vascular malformation in our study followed by LM and VM. SWS, PPV and HHT were rare syndromic variants of capillary malformation. The proportion of PROS, the rare spectrum of overgrowth disorders, including KTS and CLOVES syndrome was relatively higher in our study as compared to that of large studies on vascular anomalies. Multicentre studies are needed to study the prevalence and clinical profile of cutaneous vascular anomalies in India.

**Keywords:** vascular anomalies; vascular tumours; vascular malformations; hemangioma; overgrowth; Klippel-Trenaunay syndrome.