Abstracts

A16

Aim was not only to create simulation of real clinical situations but also to teach practical skills and build the concept of team working. ALS Manikin was modified as simple, affordable, feasible and effective (SAFE)

Methods Clinical examination, detailed family history, imaging (ultrasound, MRI, angiography) and genetic testing.

Results Patient 1 was born with a large vascular mass affecting the right side of the face and multiple cutaneous capillary malformations. Patient 2 had a spinal AV fistula and two vascular stains. Patient 3 presented with an intracranial haemorrhage secondary to a parietal AVM and was noted to have several cutaneous vascular lesions. Patients 2 and 3 were referred to the dermatology team as suspected HHT. The cutaneous vascular lesions present in all three patients were consistent with capillary malformations (in the shape of RASA-1 mutation) and were not typical of telangiectases.

Conclusion In patients with high flow CNS vascular lesions, it is crucial to establish the precise nature of cutaneous vascular lesions in order to request appropriate genetic testing and screening of relatives.

REFERENCE
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CAPILLARY MALFORMATIONS – ARTERIOVENOUS MALFORMATIONS/ARTERIOVENOUS FISTULA SYNDROME (CM-AVM SYNDROME): AN UNDER RECOGNISED CLINICAL ENTITY?

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Background Hereditary Hemorrhagic telangiectasia (HHT) tends to be the first condition to be considered in the differential diagnosis of patients presenting with high flow vascular malformations in combination with cutaneous vascular lesions. However, particularly in the paediatric population, capillary malformation-arteriovenous malformation syndrome (CM-AVM) due to RASA-1 mutation is more likely.

Aims To present the clinical features of three patients with CM-AVM syndrome, promote knowledge of this condition and aid prompt diagnosis.

Methods Clinical examination, detailed family history, imaging (ultrasound, MRI, angiography) and genetic testing.

Results Patient 1 was born with a large vascular mass affecting the right side of the face and multiple cutaneous capillary malformations. Patient 2 had a spinal AV fistula and two vascular stains. Patient 3 presented with an intracranial haemorrhage secondary to a parietal AVM and was noted to have several cutaneous vascular lesions. Patients 2 and 3 were referred to the dermatology team as suspected HHT. The cutaneous vascular lesions present in all three patients were consistent with capillary malformations (in the shape of CM-AVM) and were not typical of telangiectases.

Conclusion In patients with high flow CNS vascular lesions, it is crucial to establish the precise nature of cutaneous vascular lesions in order to request appropriate genetic testing and screening of relatives.

REFERENCE
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G25

RASA1 MUTATIONS AND VEIN OF GALEN ARTERIAL MALFORMATIONS

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