

Familial Cerebral Cavernous Malformation: case series followed at VasCog Clinic at Careggi Stroke Unit.

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INTRODUCTION

Cerebral cavernous malformations (CCMs) are rare vascular lesions affecting approximately 0.5% of the population, characterized by abnormal capillary structures in the central nervous system. Most frequently, CCMs are sporadic (90%) with single brain or spinal lesions, and often asymptomatic. A minority of cases (10%) presents familial forms (fCCM), caused by autosomal dominant mutations.

Genes involved in familial forms



CCMs can be asymptomatic or manifest with

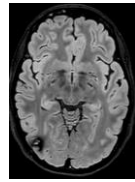


METHODS

fCCM cohort – VasCog clinic – Careggi University Hospital
Consecutive patients with a confirmed genetic diagnosis of fCCM, able to attend an outpatient visit



- Annual neurological visit
- Neuropsychological assessment



- Annual 3T brain MRI
- T2*, GRE/SWI sequences
- Lesion count and mapping

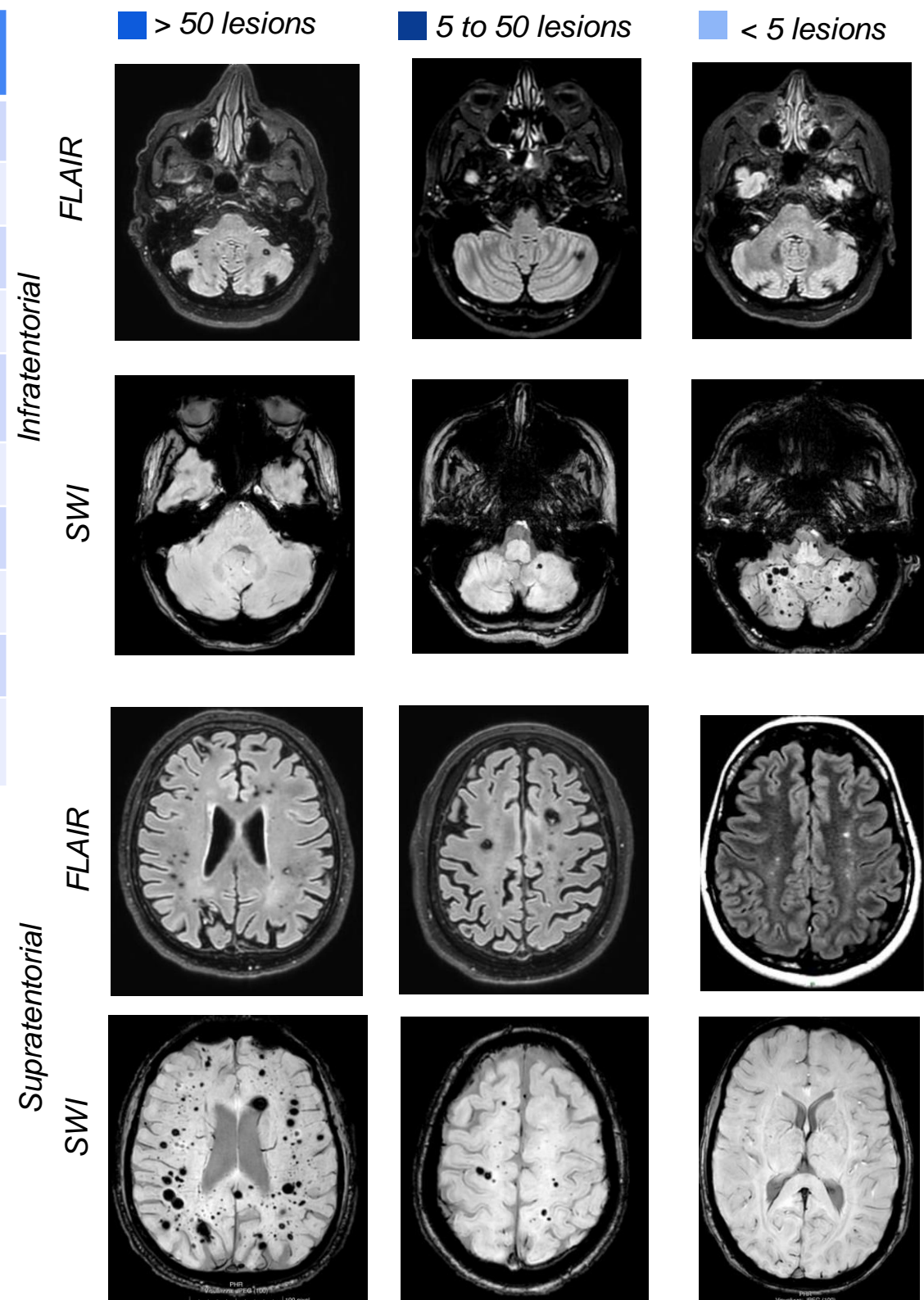


- Ultra-widefield retinography
- OCT angiography
- Retinal angioma screening

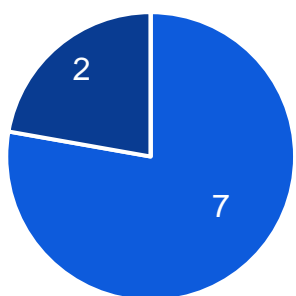
RESULTS

Pts	Age	Sex	CCM	Clinical manifestations
1	70	M	CCM1	Headache
2	64	F	CCM2	Headache
3	65	F	CCM1	Hemorrhages
4	55	M	CCM1	Headache
5	56	M	CCM1	Headache, seizure, hemorrhages
6	42	F	CCM3	Headache, seizure
7	41	F	CCM1	Headache, seizure
8	79	M	CCM1	Seizures, FND
9	34	F	CCM2	Asymptomatic

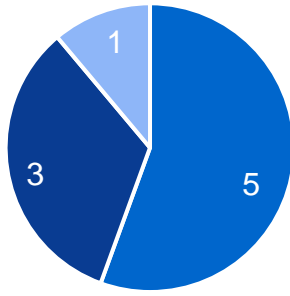
Ophthalmological assessment: none of the patients had ocular or retinal lesions



Supratentorial



Infratentorial



CONCLUSIONS

Our fCCM cohort confirms **significant clinical and radiological heterogeneity**, with KRIT1 mutations predominant (6/9) and headache as the most common symptom (67%). **Supratentorial lesions** were more frequent than infratentorial. No retinal angiomas were detected, consistent with literature data. **Multidisciplinary annual follow-up** is essential for comprehensive care. As a rare entity, larger patient registries and long-term follow up studies on fCCM are highly needed to better characterize its natural history and clinical implications.

Current collaboration with "CCM Italia"