What is the Cerebral Cavernous Malformation CCM1 Common Mutation?

Information for Patients

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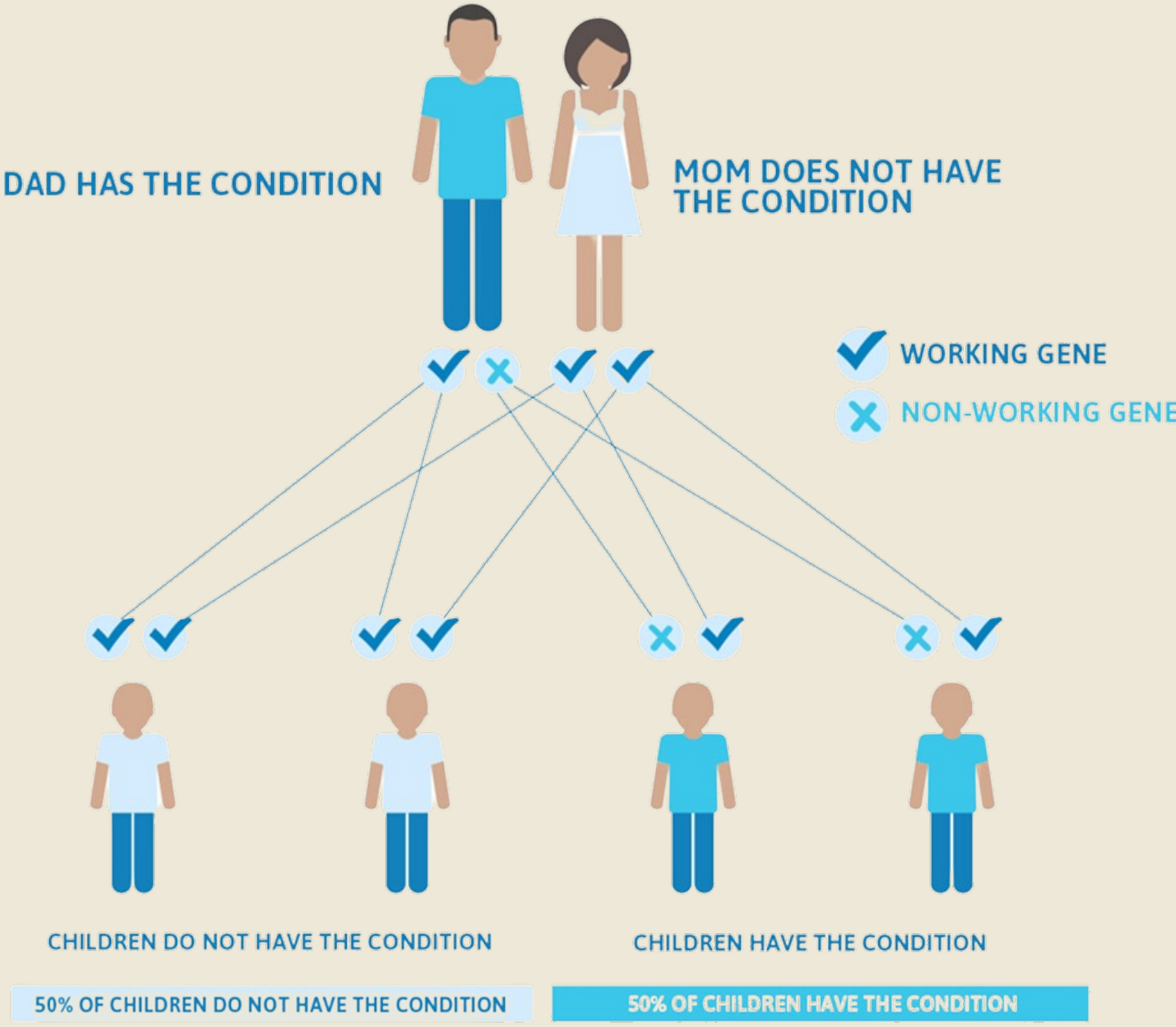
CAVERNOUS MALFORMATION

Autosomal Dominant Inheritance Pattern



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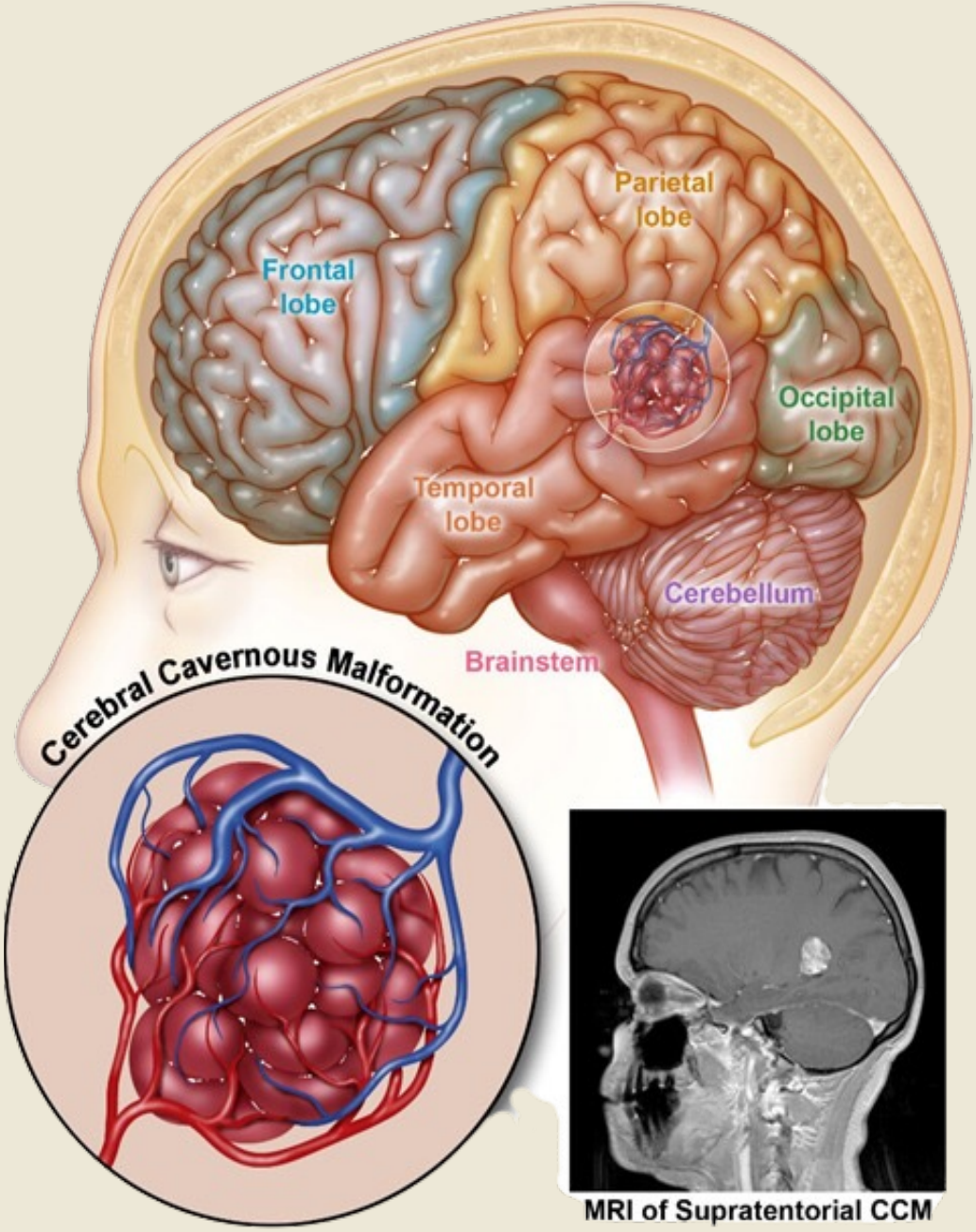
CAVERNOUS MALFORMATIONS

are mulberry-shaped, thin-walled, leaky
blood vessels with slow blood flow.



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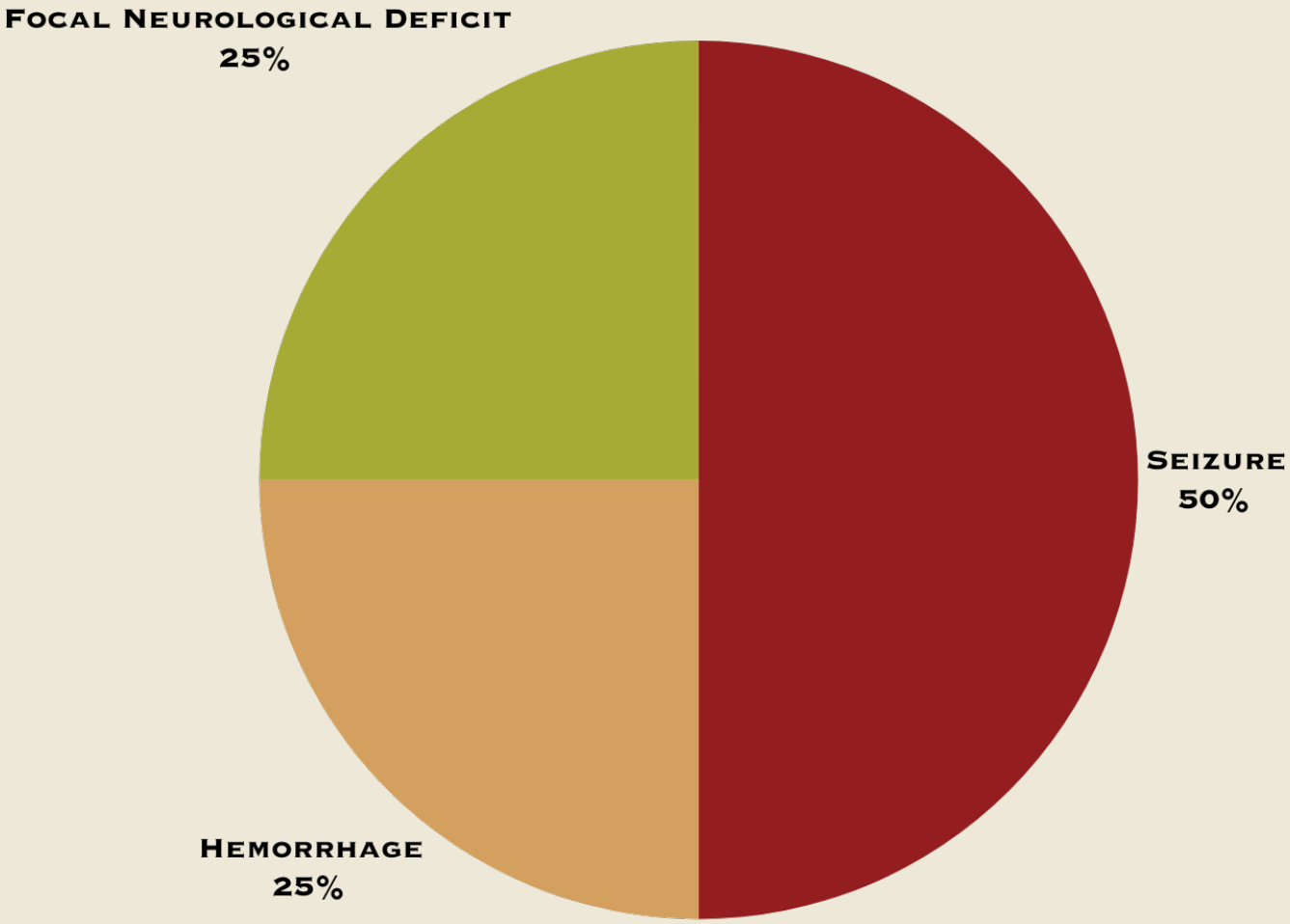
SYMPTOMS THAT LEAD TO DIAGNOSIS



A focal neurological deficit is any symptom tied to a specific area of the brain or spinal cord. Examples: arm or leg weakness, blurry vision, or facial paralysis.

At least half of those with the CCM1 Common Hispanic Mutation never have a symptom.

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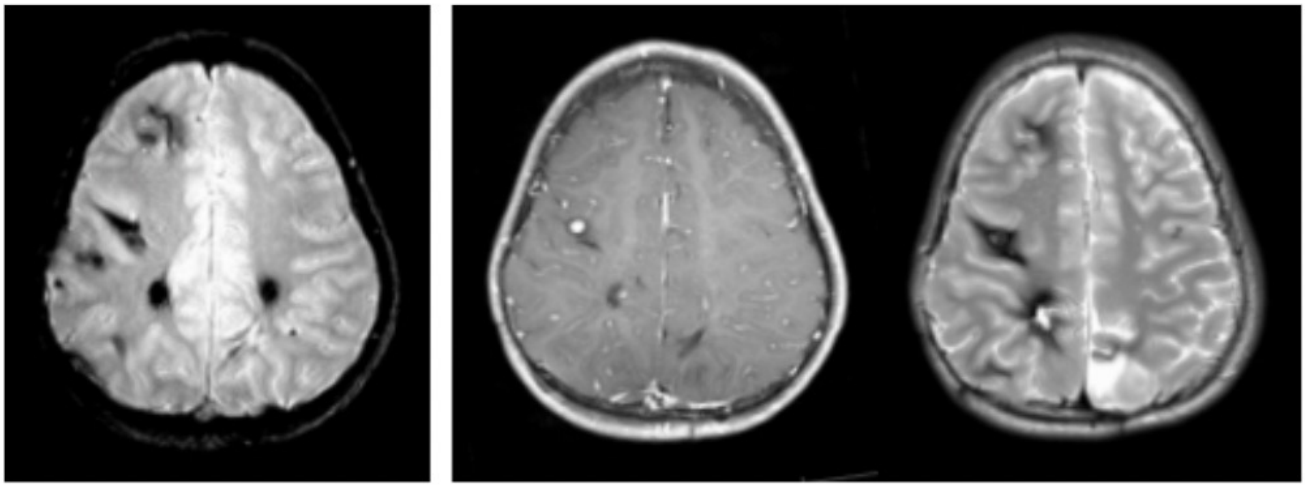
DIAGNOSING CCM: MEDICAL IMAGING



MRI with multiple cavernous malformations as seen on SWI (T2), T1, and T2 sequences.*

People who are suspected of having CCM should have Magnetic Resonance Imaging (**MRI**) for a definite diagnosis.

Cavernous malformations can look like **popcorn** with a dark ring in some scan images. With other settings, they look like dark or light spots.

POSSIBLE CCM1 SYMPTOMS

Focal seizures – uncontrolled movement in a limb or the face, smelling something that's not there.

Limb weakness, tingling, burning

Vision issues – double vision, jumpy vision, eye turning in, loss of part of visual field

Balance or coordination problems, dizziness that won't stop

Facial paralysis that resembles Bell's Palsy

Diaphragm spasms (resemble hiccups) that continue for an extended period. This is an emergency.

Projectile vomiting and loss of consciousness are also emergencies.

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Actions That May Help reduce risk of hemorrhage

Remove artificial preservatives from your diet. Use the FIG app to identify problem ingredients.

Take a Vitamin D supplement if your levels are low.

Ask for a sleep study and treat sleep apnea if you suspect you may have it.

Limit hormone replacement therapy and oral contraceptives.

Reduce inflammation: stop smoking, limit infectious disease (hand-washing, vaccination).

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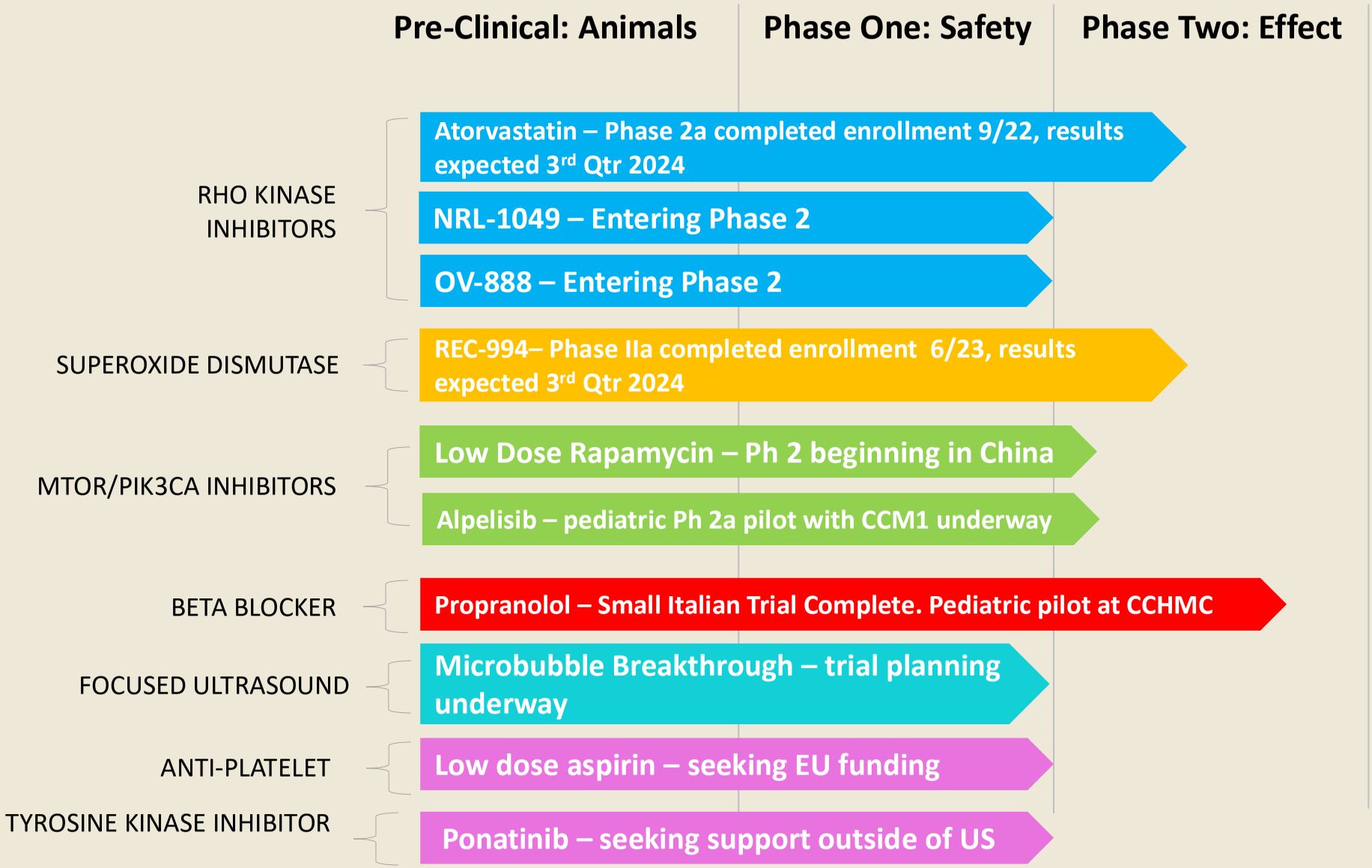
CCM Treatments in Development



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Genetic testing

How: Performed with saliva, gum swab, or blood sample. Takes about 4-6 weeks for results.

Where: Available thru UNM Neurology (505) 272-3160

Protection: Genetic Non-Discrimination Act prevents discrimination in insurance & employment.

Why test? Testing reduces the risk of misdiagnosis of symptoms.

Why test? Testing provides information to take precautions & have early access to treatments.

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