



[CCM1 GENEALOGY](#)

[CCM1 INFORMATION](#)

[CLINICAL TRIALS](#)

[RESOURCES](#)

**ALLIANCE TO CURE**  
CAVERNOUS MALFORMATION



# Clinical Trials

## PATIENT EDUCATION MATERIALS

### An Introduction to Clinical Trials

#### VIDEOS

*Videos are only available while connected to the internet unless you download them to your device.*



**Screening Appointment  
for the Rec-994 Trial**



**Rec-994 Trial Day 1**

#### MATCH TO A CLINICAL TRIAL

When you click Start below, you will be directed to the Antidote website where you will be asked for your condition and your location. You will then be asked a series of questions that will narrow your results to trials for which you might qualify. *Because this website is external to our app, you must be connected to the internet to view it.*

**Match to clinical trials in  
60 seconds**

- Know your options
- Access the latest treatments
- Receive world class care

START



# Resources

## New Mexico CCM Resources

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### **Match to clinical trials in 60 seconds**

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THE GENEALOGY  
OF THE CCM1  
COMMON  
MUTATION & THE  
BACA FAMILY IN  
NEW MEXICO



**ALLIANCE TO CURE**

CAVERNOUS MALFORMATION



# New Mexico Prevalence

In the US, 1 in 500 people have any kind of CCM, and 1 in 4000 have a CCM1 mutation.

Because of a founder mutation, in New Mexico, Hispanic residents have a higher incidence of CCM1 mutation than anywhere in the world, with concentrations along the path of the Camino Real.

Most are not diagnosed. There are between 30,000-40,000 affected individuals in the state.

High-risk names include CdeBaca, Campos, and many more.

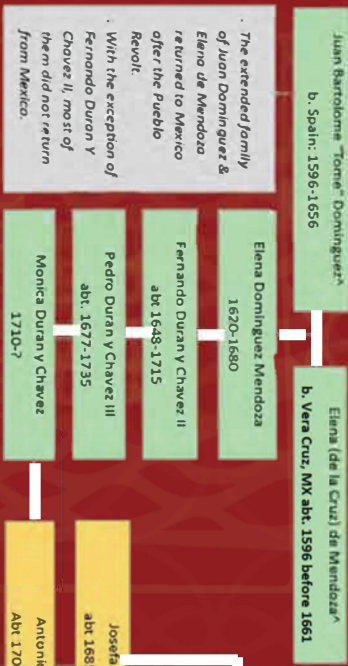
**ALLIANCE TO CURE**  
CAVERNOUS MALFORMATION



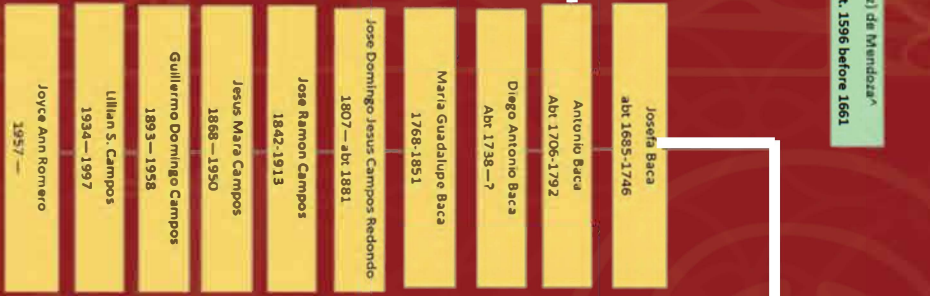
# New Mexico Prevalence Hotspots



New Mexico & Southwest Connection



- Mexico Surnames Possibly Affected by CCM1 (to date)**
- Aguirre
  - Chavez (Duran y Chavez)
  - Contieras
  - Dominguez
  - Dominguez de Mendoza
  - Grijalva
  - Mendoza
  - Perez
  - Ponce (de Leon)
  - Rascón
  - Véjar



- New Mexican Surnames Possibly Affected by CCM1 (to date)**
- |              |                         |            |           |
|--------------|-------------------------|------------|-----------|
| Apodaca      | Chavez (Duran y Chavez) | Luna       | Rivera    |
| Aragon       | Espinosa/Espinoza       | Maes/Maesé | Romero    |
| Arimo        | Gallegos                | Martinez   | Sandoval  |
| Baca         | Garcia                  | Mora       | Silva     |
| * C' de Baca | Gonzales                | Ortiz      | Saiz/Saiz |
| Campos       | Lopez                   | Padilla    | Vigil     |
| Casaus       | Lucero                  | Quintana   |           |

<sup>a</sup>From the same family that lived in the area of the present-day state of Veracruz, Mexico.

THROUGH GENEALOGY, WE CONTINUE TO DISCOVER  
 MORE SURNAMES AND FAMILY LINES THAT MIGHT BE AT  
 HIGHER RISK FOR THE CCM1 COMMON MUTATION.

THE SURNAMES WE KNOW HAVE A HIGHER INCIDENCE OF  
 THE CCM1 COMMON MUTATION IN NEW MEXICO ARE:

APODACA	CHAVEZ (DYC)	MAES/MAIS	ROMERO
ARAGON	ESPINOSA/ZA	MARTINEZ	SANDOVAL
ARMIJO	GARCIA	MORA	SILVA
BACA	GONZALES	ORTIZ	SAIS/SAIZ
C' DE BACA	LOPEZ	PADILLA	VIGIL
CAMPOS	LUCERO	QUINTANA	
CASAUS	LUNA	RIVERA	



THROUGH GENEALOGY, WE CONTINUE TO DISCOVER MORE  
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THE SURNAMES THAT WE KNOW HAVE A HIGHER INCIDENCE  
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AGUIRRE

CHAVEZ

CONTRERAS

DOMINGUEZ

DOMINGUEZ DE MENDOZA

CAMPOS

CONTRERAS

GRIJALVA

MENDOZA

PEREZ

PONCE (DE LEON)

RASCON

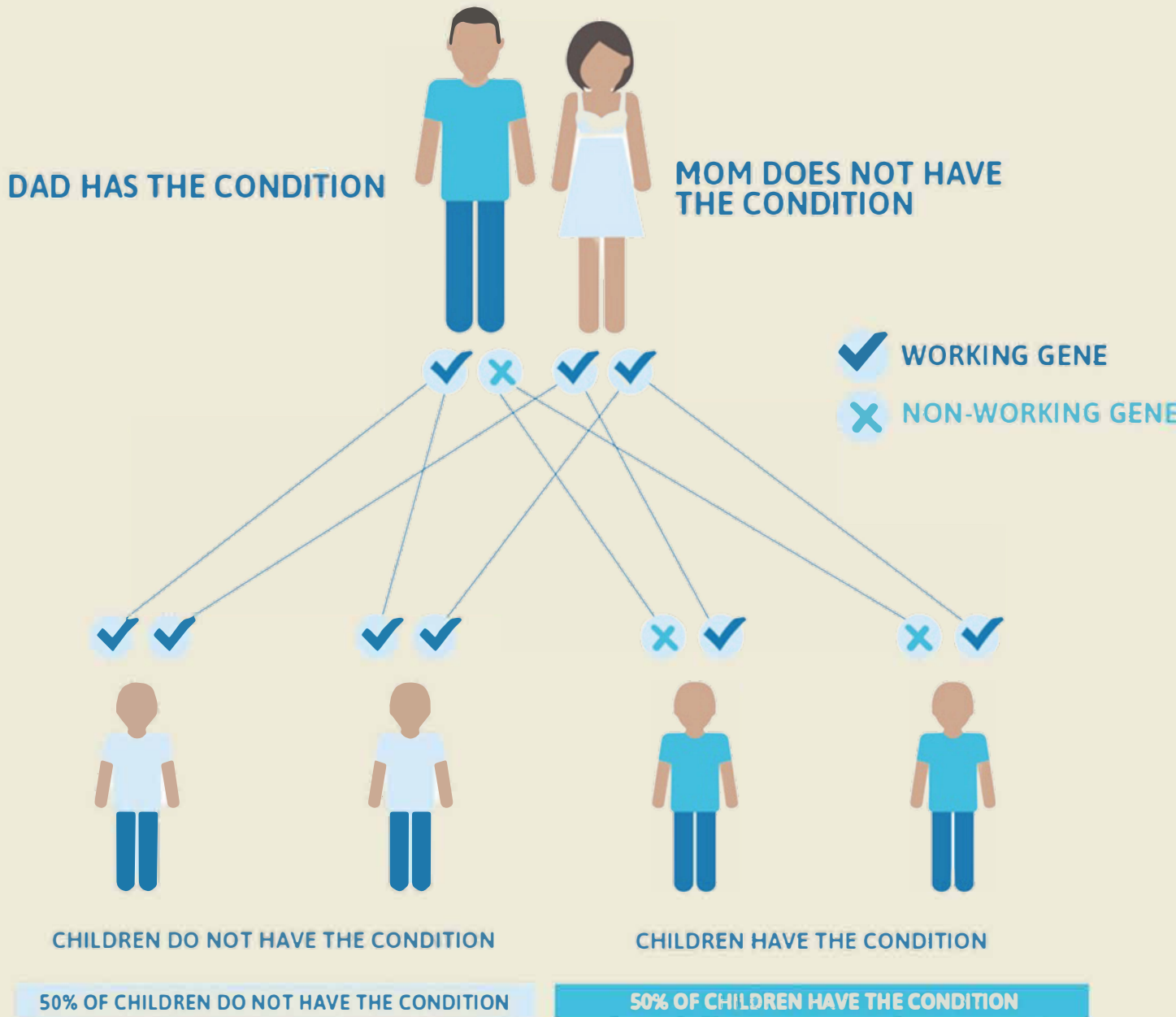
VIGIL

# **What is the Cerebral Cavernous Malformation CCM1 Common Mutation?**

## **Information for Patients**

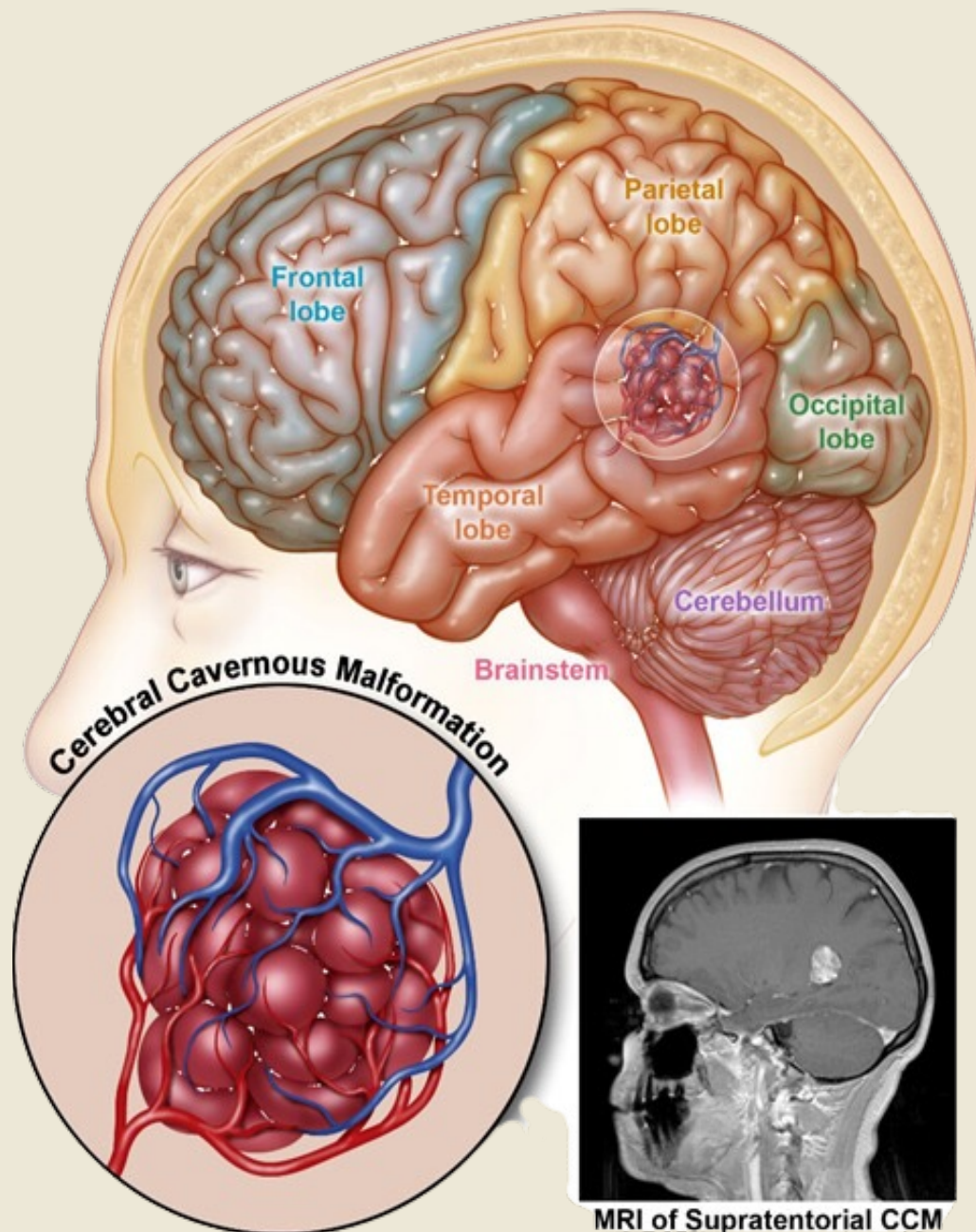


# Autosomal Dominant Inheritance Pattern



# CAVERNOUS MALFORMATIONS

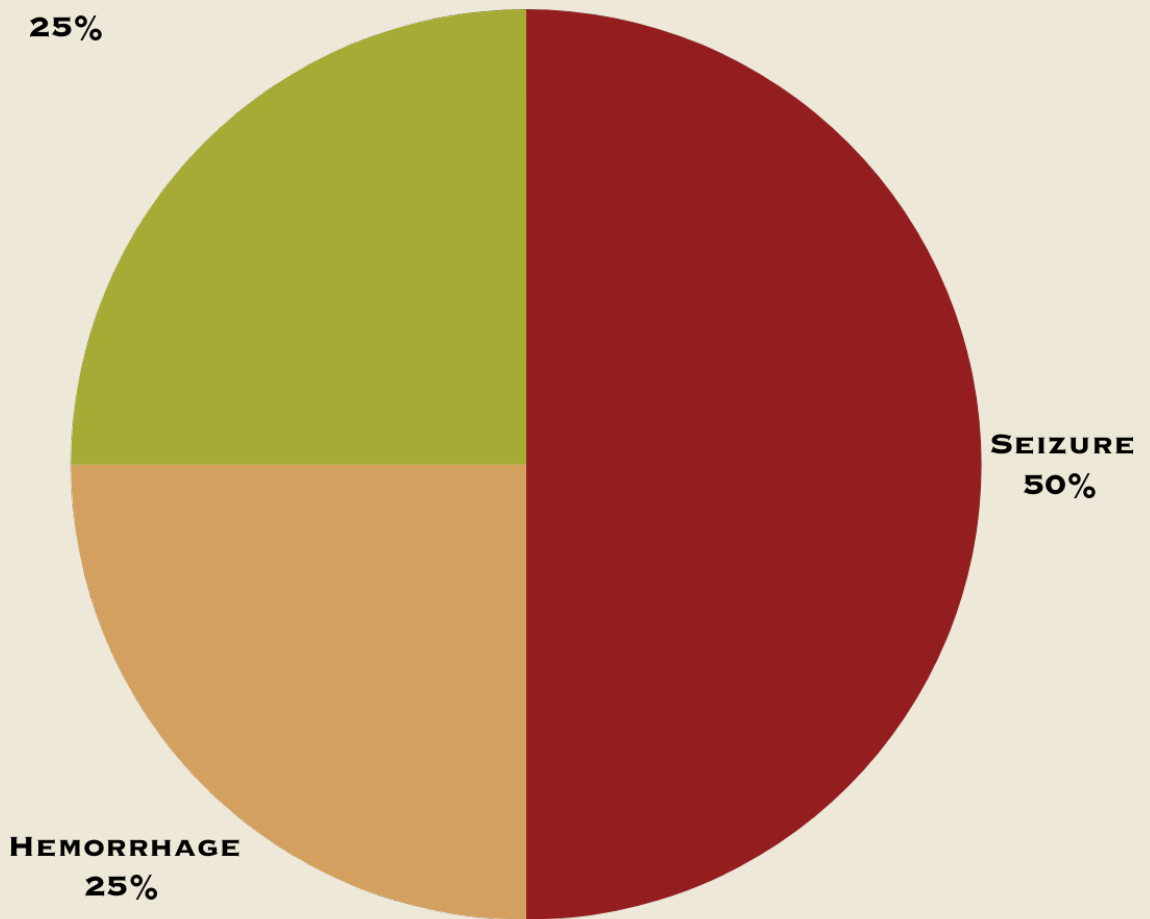
are mulberry-shaped, thin-walled, leaky blood vessels with slow blood flow.



# SYMPTOMS THAT LEAD TO DIAGNOSIS

**FOCAL NEUROLOGICAL DEFICIT**

**25%**



**SEIZURE**  
**50%**

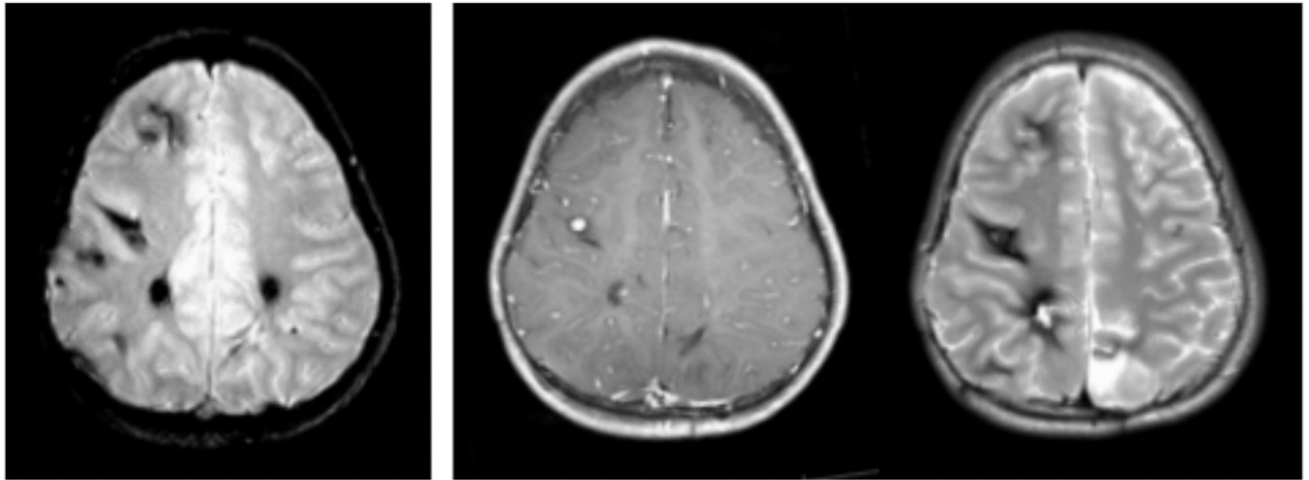
**HEMORRHAGE**  
**25%**

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.



# 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.

# 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).



# CCM Treatments in Development



# 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.

# An Introduction to CLINICAL TRIALS



# CLINICAL TRIALS

## ALLOW US TO LEARN:



WHETHER A NEW TREATMENT IS  
SAFE



WHETHER A NEW TREATMENT IS  
EFFECTIVE OR MORE EFFECTIVE FOR  
SOME PEOPLE THAN OTHERS.



HOW MUCH OF A NEW TREATMENT  
IS NEEDED TO HAVE THE DESIRED  
EFFECT – THE DOSE & FREQUENCY

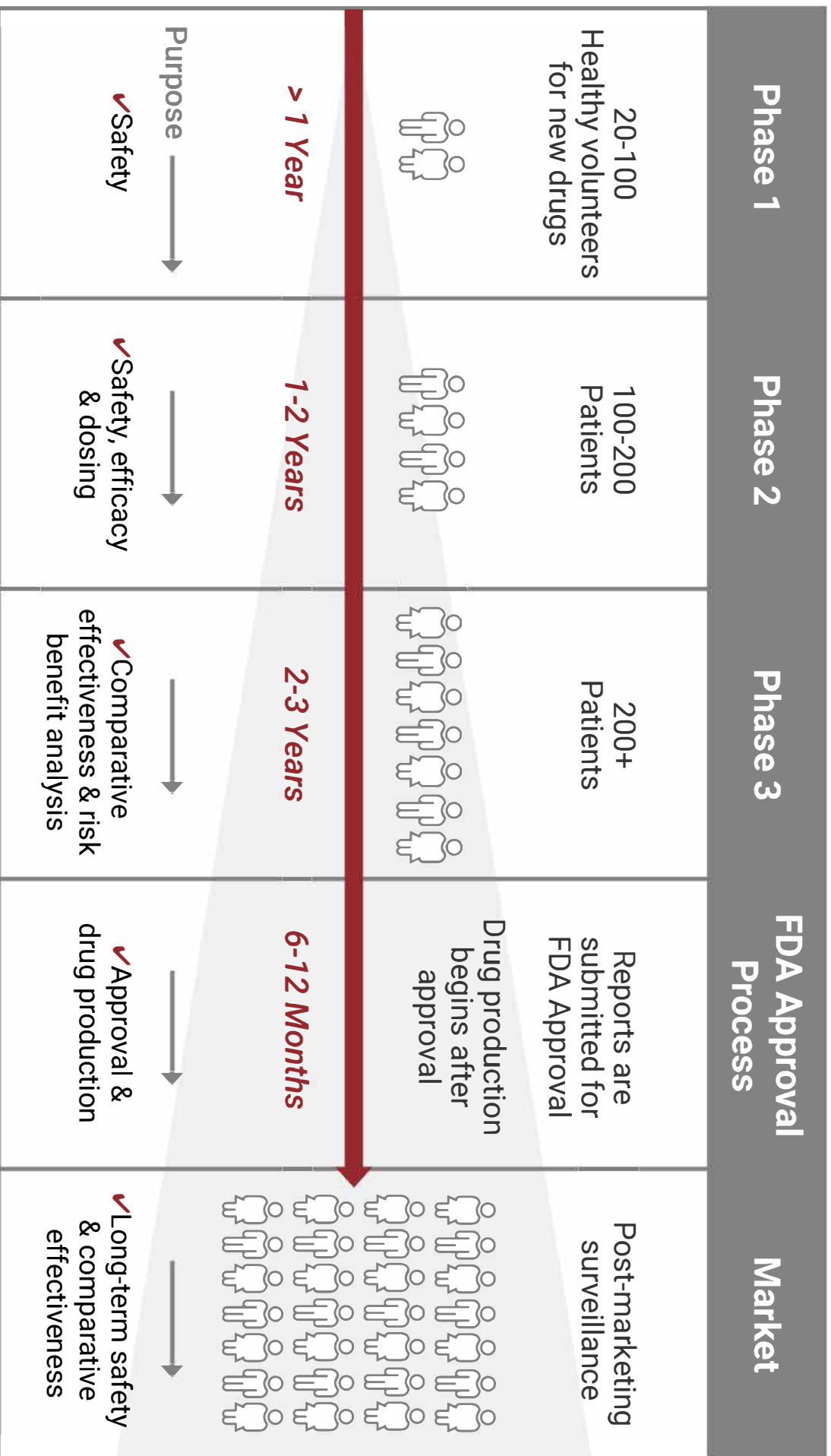


WHETHER A NEW TREATMENT HAS  
UNINTENDED SIDE EFFECTS.



# Clinical Trials

## The Drug Development Process



The duration of a trial and required number of participants is dependent upon the outcome that is measured. Trials that measure hemorrhage, for example, an infrequent event, are likely to be longer in duration than a trait that measures a more frequent outcome, like headache or seizure.

# PLACEBOS



## “Placebo effect”

A placebo is a substance or procedure that appears identical to the treatment being tested.

Sometimes, a condition will improve just because a person thinks they are receiving treatment.



Some participants in larger clinical trials will take a placebo to ensure the real treatment is better. In trials where it would be unethical to withhold treatment, placebos are not used.

# WHAT IS THE BENEFIT OF PARTICIPATING IN A CLINICAL TRIAL?

The investigational treatment studied in a clinical trial may or may not benefit the patient personally. The benefits of participating in a clinical trial may include:

## CONTRIBUTION

Helping other patients by contributing to medical research and treatment advances.

## ACCESS

Gaining access to cutting-edge research.

## CARE

Receiving expert medical consultation for the condition being studied, since doctors conducting clinical trials are often specialists in the disease areas.

# MYTHS VERSUS FACTS

Here are some common myths about clinical trials and the facts.

## I CAN'T DROP OUT

TRUTH: You may leave a trial at any time, for any reason.

## IT COSTS TO PARTICIPATE

TRUTH: Most trials are free. Participant's travel is reimbursed, and they receive a stipend.

## I MUST STOP ALL OTHER MEDICINES

TRUTH: Every trial has different requirements. Many allow other medicines.

## I MUST CHANGE DOCTORS

TRUTH: You can keep your current doctor for care and use the trial doctor for trial monitoring.



# HOW ARE CLINICAL TRIAL PARTICIPANTS KEPT SAFE?



EACH TRIAL IS MONITORED BY MEDICAL STAFF, AND PARTICIPANTS ARE REGULARLY SEEN.



HEALTHY VOLUNTEERS HAVE ALREADY TAKEN THE TREATMENT AND NOT HAD SERIOUS SIDE EFFECTS.



EACH TRIAL IS APPROVED & OVERSEEN BY MULTIPLE SAFETY AUTHORITIES: FDA, INSTITUTIONAL REVIEW BOARD, DATA SAFETY MONITORING BOARD.



THE TRIAL MUST PROVIDE YOU WITH ALL THE INFORMATION YOU NEED TO MAKE A DECISION ABOUT PARTICIPATING.

# 5 Phases of Decision-Making

Deciding to be part of a clinical trial is a progression and the messages can be mapped to this progression.

## 1. Precontemplation

- Has never heard of a clinical trial
- Is not interested in participating

## 2. Contemplation

- Knows some about clinical research
- Is willing to learn more to consider participation

## 3. Preparation

- Made an appointment with researchers
- Is asking questions about participating

## 4. Action

- Has been pre-screened by PCP and PI
- Has read consent form

## 5. Maintenance

- Has signed consent form
- Is not a screen failure
- Has come to baseline appointment

These are the five phases of decision making in the transtheoretical model.

\* PI = Principal Investigator. This is the doctor in charge of the clinical trial at the clinical trial site.

## **New Mexico CCM1 Resources**

### **Clinical Care**

The **University of New Mexico Health System** has been designated a CCM Center of Excellence by the Alliance to Cure Cavernous Malformation.

As with most UNM departments, the demand for services is often greater than the number of available appointments, so wait times to get an appointment can be long.

However, UNM Neurology, Neurosurgery, and Genetics departments are staffed with CCM1 experts who have seen more patients than any facility in the world. UNM Neurology has been funded by the State to offer free genetic testing.

Patients also have multiple opportunities to participate in research, including upcoming clinical trials. UNM hosts an annual patient conference, typically in the spring.

Appointment line: 505-272-3160

## **UNM CCM Center of Excellence Faculty**

**Medical Director and Vascular Neurologist:** [Dr. Michel Torbey](#)

**Cerebrovascular Neurosurgeons:** Dr. Andrew Carlson, Dr. Anish Deshmukh, and Sean Deloach, NP

**Vascular Neurologists:** Dr. Tarun Girotra, Dr. Tobias Kulik, Dr. Monika Manchanda, Dr. Maryam Hosseini

**Epileptologist:** Dr. Jose Padin-Rosado, Dr. Ken Imerman, Dr. Anna Bhat

**Geneticist:** Dr. Randall Heidenreich

**Pediatric Neurology:** Dr. Kathy Wolfe

**Genetic Counselor:** Joanne Drautz, CGC

**Neuroradiology:** Dr. Mark Mabray

**Nurse Coordinator:** Dawn Aldridge, RN

**Nurse Educator –** Michael Richardson, RN

**Educator Coordinator –** Brittany Gagne

**UNM Research Program Patient Enrollment:  
505-272-3194**

### **Patient Support**

New Mexico hosts an active **chapter of the Alliance to Cure Cavernous Malformation**. The group hosts regular gatherings and awareness events. To connect with the group, please contact Linda Fuchser at [linda@alliancetocure.org](mailto:linda@alliancetocure.org).