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MADISON NEUROLOGY LLC

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Neurology Care 7 days a Week. Because Your Brain Matters.

CADASIL (Inherited Small Vessel Disease)

Patient education guide · Stroke & Vascular · Madison Neurology LLC

What is it?

CADASIL (cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy) is an inherited disorder of the brain's small blood vessels caused by a mutation in the NOTCH3 gene. It leads to migraine with aura (often beginning in the 20s-30s), recurrent small strokes (typically from the 40s-50s), mood changes, and gradually progressive cognitive decline.

How does it happen?

CADASIL is caused by mutations in the NOTCH3 gene and is passed in an autosomal dominant pattern — each child of an affected parent has a 50% chance of inheriting it. The abnormal protein damages the muscle layer of small arteries, impairing brain blood flow.

Common symptoms

- Migraine with aura, often starting unusually early or with prolonged auras
- Recurrent small (lacunar) strokes or TIAs
- Depression, apathy, or other mood changes
- Gradual problems with memory, processing speed, and executive function
- Walking difficulties and urinary problems in later stages

How is it diagnosed?

Brain MRI shows a characteristic pattern of small-vessel changes, including lesions in the anterior temporal poles — a strong clue. Genetic testing for NOTCH3 mutations confirms the diagnosis; skin biopsy is an older alternative.

How is it treated?

There is no disease-specific cure yet, so care focuses on vascular risk control: strict blood pressure management, not smoking, statins when indicated, and standard stroke prevention (though routine blood thinners are individualized). Migraine is treated with standard preventive and acute therapies, avoiding triptans only per specialist guidance. Genetic counseling is essential for family planning.

What does the future hold?

The course is slowly progressive over decades, but varies considerably even within families. Many people remain active for many years after diagnosis; dementia and disability typically develop later in life. Research into disease-modifying treatments is active, and registries connect families with studies.

Questions patients commonly ask

Q: Should my relatives be tested?

A: Adult relatives can choose predictive genetic testing after counseling; testing is a personal decision and should be guided by a genetics professional.

Q: Can I still take triptans for migraine?

A: This is debated; many specialists permit them in CADASIL, but the decision should be made with your neurologist.

Q: Does everyone with the gene get sick?

A: Yes, essentially everyone with a CADASIL-causing NOTCH3 mutation develops signs over time, but severity and timing vary widely.

National support groups & resources

- **American Stroke Association** — stroke.org — 1-888-478-7653
- **National Organization for Rare Disorders** — rarediseases.org — 1-800-999-6673
- **American Academy of Neurology — Brain & Life patient resources** — brainandlife.org

(Organization contact details were current at the time of compilation; please verify online.)

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