



<https://ca2deficiency.duckdns.org/> · Educational organizing tool — not a diagnosis or treatment plan

Genetic Counseling and Pregnancy Planning

General information only. Does not advise continuing or ending a pregnancy.

- Autosomal recessive inheritance is the usual genetic framework for this condition.
- Confirmed family variants matter when available.
- Bring genetic reports to counseling.
- Discuss recurrence risk with a genetics team.
- Prenatal testing options may exist.
- PGT may be discussed when available.
- Referral to genetics and/or maternal-fetal medicine.

Genetic reports we have (families need not paste full results)

Questions for the team

Any prenatal diagnosis and reproductive decision should be discussed with qualified genetics and maternal-fetal medicine teams according to applicable medical, ethical, religious, and legal frameworks.

Educational and organizational only. Does not diagnose, set doses, or replace a clinician. What you notice at home is not the same as what is in the medical record, and not the same as what the doctor decides.