MTHFR Fertility

FERTILITY CASE STUDY

RECURRENT EARLY PREGNANCY LOSS

Hello and welcome!

I'm Carolyn Ledowsky, founder of **MTHFR Fertility and MTHFR Support Australia.**

As an MTHFR researcher, trainer and presenter, I am committed to teaching everyone I can about how and why the MTHFR genetic polymorphisms may affect your ability to fall and stay pregnant. I find it so sad that women are needlessly having multiple miscarriages and suffer the heartache that goes along with that, yet if they had known they had the MTHFR gene mutation before they started out they would have done things differently.

In honour of **National Infertility Awareness Week,** my team and I have compiled some amazing case studies and resources to share with you.

Our goal is to **support, inspire and empowe**r everyone going through this sometimes heartbreaking journey, to find the answers (and hope!) they need to turn things around.

Enjoy!

Carolyn Ledowsky

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Case Study:

Recurrent Early Pregnancy Loss

The following is a real-life case study of a couple who had been trying to conceive for 2 years. They had experienced 4 miscarriages.

Ages

- Female 31
- Male 31

Presenting signs and symptoms (female)

- Trying to conceive 2 years
- 3 miscarriages @ 9 weeks
- 1 miscarriage @ 13 weeks
- no autoimmune markers or previous health conditions
- Both currently taking a prenatal with 500mcg folic acid, female taking extra 500mcg folinic

Medical Diagnosis + Advice

- Bad luck
- Keep trying

Further Testing Revealed...

- Low homocysteine indicating methylation disturbance
- Anaemia caused by low folate, low B12 and low iron
- low vitamin D
- chronically low white blood cells (indicating chronic infection or B12 deficiency)

MTHFR Fertility

What genes can affect Miscarriage?

Genes We Assess for Miscarriage

- Folate Genes e.g. MTHFR, MTHFD1, MTHFD1L, DHFR, SHMT, TYMS, SLC19A1
- B12 Genes e.g MTR, MTRR,TCN1, TCN2, FUT2
- Choline e.g. PEMT, BHMT, DMGDH

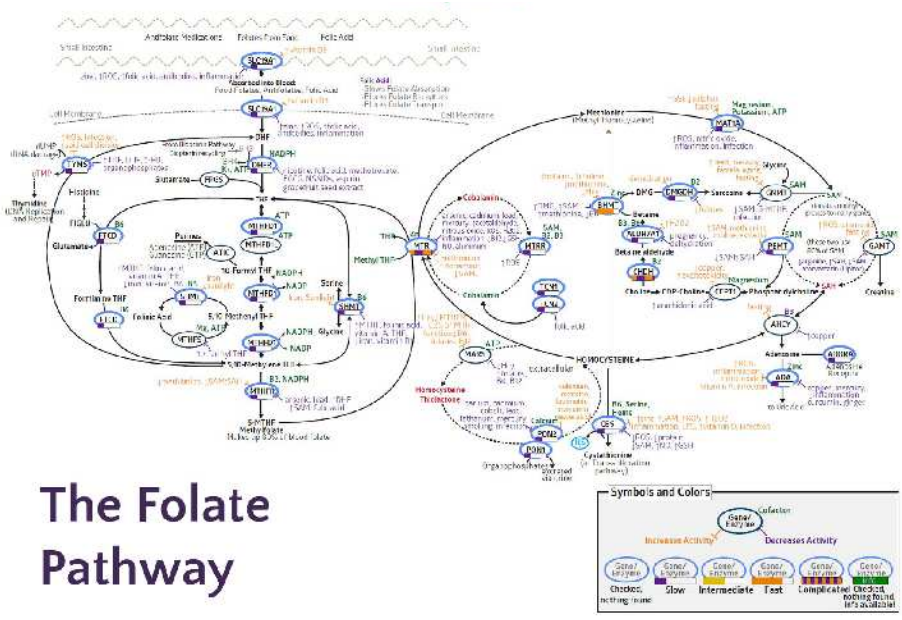
Case Study Genetics Discussion

- **Folate, B12 + Methylation Pathway: C667T++ (female) Compund Heterozygous (male)**

Both partners showed a significant reduction in MTHFR activity (70%- 50% respectively). Both also presented with DHFR ++ (at the top of the folate pathway) and MTHFD1. Folate and B12 deficiency was evident in both partners. SHMT mutations also meant folate recycling was reduced indicating a higher need for B6.

Female also presented with a TCN1 mutation which can cause reduced absorption of B12 in the gut and cause B12 deficiency. There was initially no B12 supplementation other than the 2 mg found in the prenatal.

Getting the right type of Folate and B12 was essential for both partners. Remember folate is essential for the health of DNA within the egg AND sperm. Both partners needed high-dose methyl folate and methylcobalamin (B12).

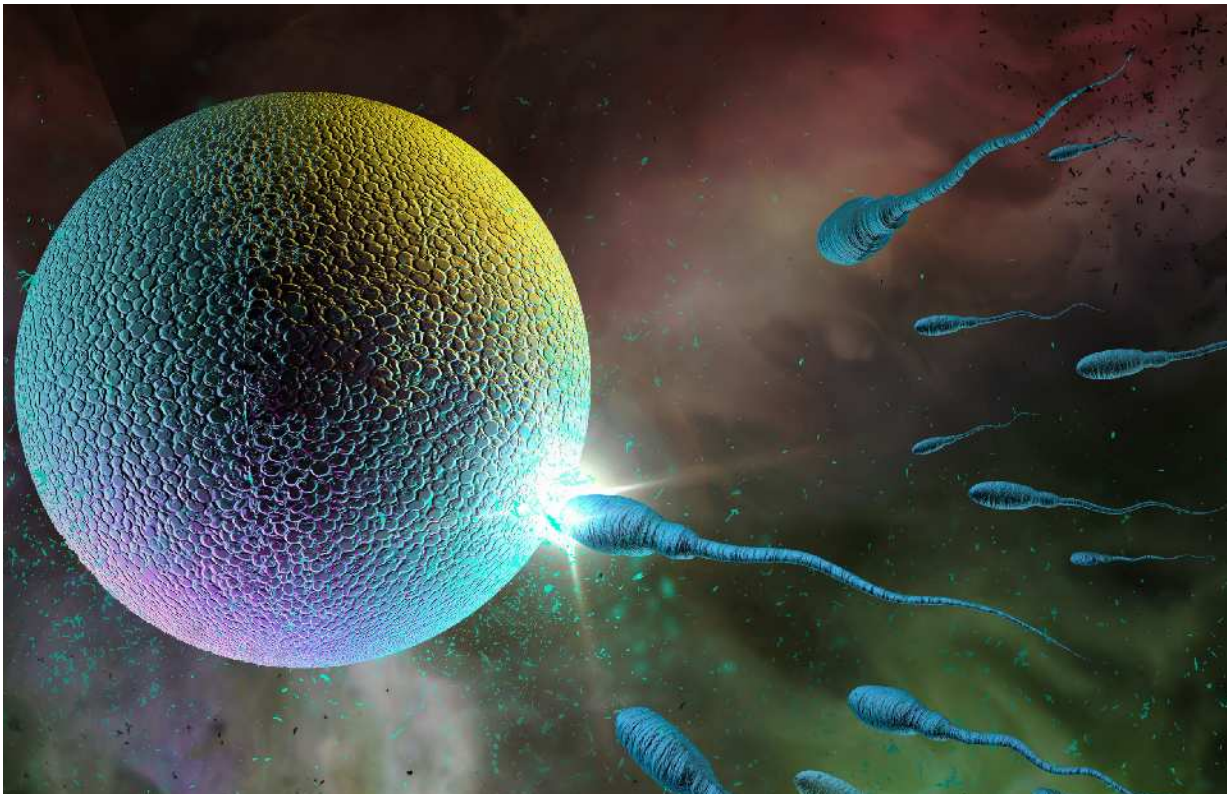


Key Genetics Continued...

- Choline

The choline genes/ enzymes can act as a "back-up" to methylation and get up-regulated when the folate pathway is slowed. Choline is also essential for healthy cell membranes that protect the DNA within the sperm and eggs.

Hence, Insufficient choline will impact the health of the sperm and eggs. The female had multiple genetics that slowed the production of "active" choline needed for healthy cell membranes. Including PEMT, BHMT and DMGDH. She was not currently taking any supplements.



MTHFR Fertility

Treatment Overview

Both partners

Step 1: Support Methylation

- Switch to MTHFR-suited prenatal (no folic acid)
- Add additional high-dose methyl folate
- Methyl B12 supplements
- Phosphatidylcholine
- B6 (female)

Step 2: Additional Nutrients

- Iron (female)
- Innositol (female)
- N-acetyl-cysteine (both)

RESULTS - Pregnant within 1.5 months

- Folate dose increased further for first trimester as the ideal 3 month folate dose was not achieved in preconception
- Healthy pregnancy

Beautiful, healthy Baby girl!

Tell Us Your Story

Have you experienced Recurrent Early Loss?

Tell us about your story in our private Facebook group or our Instagram page and use the hashtag ***#thisismystory***