

## 13. Spina bifida and factor V Leiden and other thrombophilias

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**Q: "I would be interested to know if there is a connection that a mother carrying inherited thrombophilia (factor V Leiden) has a chance of creating a spina bifida baby. The reason why I ask is, that 2 sisters in my family have inherited thrombophilia and both have spina bifida children. They both even had miscarriages too."**

A: There is no known connection between inherited thrombophilia, such as factor V Leiden, in the mother and spina bifida in the baby.

Spina bifida occurs in approximately 1 of every 2000 births. Since factor V Leiden occurs in approximately 5 % of the US population, 1 of 20 children with spina bifida will have a mother who has factor V Leiden. Thus, this is not an unusual occurrence. And since spina bifida can run in families and factor V Leiden runs in families, it is not unusual to find both in several family members. It does not mean that there is a causative connection. And indeed, there is no known connection between inherited thrombophilia and spina bifida.

Spina bifida is caused by environmental and genetic factors. The most well-known environmental factor is folate deficiency in pregnant women. Folate deficiency can lead to elevated homocysteine levels, particularly in the individual who has the homozygous MTHFR C677T mutation ( $\equiv$  thermolabile MTHFR). Since there have been some studies indicating that elevated homocysteine levels in the unborn may be associated with spina bifida, researchers have looked, whether babies with spina bifida have a higher rate of the homozygous C677T MTHFR mutation than babies without spina bifida. Some studies have found an association, others not. It is fair to conclude, that it is not clear whether the MTHFR C677T mutation in the baby plays a role in the development of spina bifida.