Homozygosis mutation of factor V Leiden revealed by a neonatal stroke

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INTRODUCTION:

Factor V Leiden mutation is the most common inherited prothrombotic factor. It's implication on venous thrombotic events has been well established. Several reports, especially in children, had described arterial ischemic events associated to this coagulation abnormality even though studies failed to establish this association. Our case is an additional report describing this association.

CASE REPORT:

- A female full term newborn was born by c-section with Apgar scores of 9 at 1 min and 10 at 5 min.
- Admitted for repeated episodes of **cyanosis** at 16 hour of life
- **Electroencephalogram**: **focal epileptic seizures** with right rolandic rhythmic spikes (figure 1).
- **Magnetic resonance imaging of the brain associated to angiogram**: recent right middle cerebral artery **ischemic stroke** (figure 2) with no signs of dissection or abnormalities of the carotid arteries.
- **Echocardiography**: normal
- **Coagulation studies**: **homozygosis G1691A mutation** of factor V Leiden.
- At 1 year old her clinical examination is normal after physiotherapy

Figure1: focal epileptic seizures with right rolandic rhythmic spikes

Figure2: MRI right middle cerebral artery stroke (diffusion tensor imaging)

COMMENTS:

- The link between activated protein C resistance (APC-R) and venous thrombosis is well established.
- The impact of factor V Leiden is age dependent and is highest in the newborn and young infants.
- In contrast to findings in adults, most of the thrombotic events associated with APC-R reported in the young are of arterial origin.
- Several studies have reported an increased frequency of factor V Leiden in childhood arterial ischemic stroke (16 to 20%)
- Anticoagulant therapy in case of cerebral thrombosis due to factor V Leiden is controversial

CONCLUSIONS:

Inherited thrombotic factor especially factor V Leiden mutation and MTHFR mutation should be kept in mind in case of arterial ischemic event on childhood.