PERINATAL THROMBOSIS AND INHERITED PROTHROMBOTIC DISORDERS

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Introduction: Prothrombotic disorders may be associated with perinatal stroke (PT) and influence outcome. An important inherited disorder is factor V Leiden mutation. PT is an acute neurologic syndrome due to cerebral injury of vascular origin, occurring between 20 weeks gestation and 28 days postnatal life. Estimated yearly incidence is 35 per 100000 live births.

Material and methods: Retrospective study of infants with prothrombotic disorders causing PT born in Hospital Cuf Descobertas between 2003 and 2011.

Results: Between 2003 and 2011, 23414 babies were born. PT occurred in 8 newborns. Coagulation disorders, factor V Leiden, have been identified in two cases, 25%.

<table>
<thead>
<tr>
<th>Stroke /Timing</th>
<th>Stroke / Type</th>
<th>Development</th>
</tr>
</thead>
<tbody>
<tr>
<td>Neonatal</td>
<td>Ischemic</td>
<td>9 Years - Normal</td>
</tr>
<tr>
<td>Late neonatal</td>
<td>Thrombosis</td>
<td>8 Years-Epilepsy, Development delay</td>
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<tr>
<td>Neonatal</td>
<td>Hemorrhagic</td>
<td>2 years –spastic diplegia</td>
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<tr>
<td>Neonatal</td>
<td>Ischemic</td>
<td>20 M - Normal</td>
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<tr>
<td>Neonatal</td>
<td>Ischemic</td>
<td>18 M – Normal</td>
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</tbody>
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Etiology

- Factor V Leiden: 25%
- Sepsis: 13%
- Thrombocitopenia: 12%
- Unknown cause: 50%

Review of the cases with Factor V Leiden

Case 1
Thrombosis detected in neonatal period.
- Vaccum delivery at 40 weeks, Newborn female, 3015 g, Apgar index 9-10.
- Started seizures 30 minutes after birth.
- MRI showed cerebral venous thrombosis of great cerebral vein of Galen and cavernous sinus.
- Factor V Leiden was identified.
- The child is presently 3 years old. Neurodevelopment is normal.

Case 2
Ischemic stroke detected in uterus.
- Elective caesarean was performed at 37 weeks. Newborn male, 3065 g. Apgar index 9-10
- D1 MRI revealed massive infarction of right middle cerebral artery.
- Factor V Leiden was identified.
- The child is now 12 months old, has left hemiparesis and normal cognitive development.

Conclusions: Our incidence of PT is similar to other series (34 /100 000 live births). Half of the cases remain without diagnosis. Inherited prothrombotic disease were identified in 25%. Since these disorders represent risk factors for PT, diagnostic tests should be performed before pregnancy, in clinically suspicious cases. Ultrasound was the first image method used. MRI define the type, extent, vascular territory and number of lesions. Long-term follow-up is necessary since neurodevelopment sequelae may be noticeable over time.