Evaluation of Anti Mullerian Hormone (AMH) assay Roche® on umbilical cord blood: Determination of reference values in newborn girls and boys.

D.Vasseur, C.Kolenda, JV Reynaud, Y.Morel, I.Plotton,
Hospices Civils de Lyon and Université Claude Bernard de Lyon, Claude Bernard University, INSERM 1208

**Background**

AMH concentration is now well studied for prepubertal boys (Plotton 2009, 2012) or women in reproductive medicine (Anderson, 2015). AMH reference values have been determined with the Roche ® automated technique, but we dispose very is few data about AMH reference value for the newborns. These value could be helpful for the management in neonatal period of neonate cases with Disorders of Sex Development (DSD).

**Objective**

Determine AMH reference values on umbilical cord blood for girls and boys neonates.

**Methods**

This study of AMH values on umbilical cord blood was realized after approval of the ethics committee between May 13th, 2015 and February 4th, 2016.

AMH assay was realized by using an automated Electro-chemiluminescence Immunoassay (Cobas Roche®).

We analyzes AMH on Heparin syringes for blood gas systematically collected after births. Personal history and clinical data for all the sample were collected simultaneously at the systematic examination (3d day or before the exit): gestational age and history of syndromic malformations or urogenital anomalies like cryptorchidism, hypospadias or micropenis diagnosed on the clinical exam. We excluded samples of neonates presenting genitourinary abnormalities.

During this period, we also evaluates AMH in 7 cases of DSD neonatal (hypospadias with intrascrotal or inguinal gonad) for which we identify an molecular explanation.

**Results**

□ 230 blood samples of venous umbilical from neonates for which no clinical abnormalities were include. This study - The samples of 192 boys and 38 girls allowed of reference interval for AMH 20 the first hours of life (See Table and graph)

<table>
<thead>
<tr>
<th>pmol/L</th>
<th>n</th>
<th>Median</th>
<th>1st Quartile</th>
<th>3rd Quartile</th>
<th>min-max</th>
</tr>
</thead>
<tbody>
<tr>
<td>Boys</td>
<td>192</td>
<td>249.95</td>
<td>178.7</td>
<td>338</td>
<td>58.5-724.3</td>
</tr>
<tr>
<td>Girls</td>
<td>38</td>
<td>1.03</td>
<td>0.56</td>
<td>2.59</td>
<td>0.28-32.5</td>
</tr>
</tbody>
</table>

AMH reference values between boys and girls significantly different (p<0.0001)

□ Among the 7 neonates, we identified:
- 3 cases of DSD with defective androgen (DA) action (mutation Androgen Receptor mutation (1), of 5 alpha reductase (2))
- 4 cases with testicular dysgenesis (TD): 1 SF1 mutation case, 3 mosaicism (45X, 46XY)

<table>
<thead>
<tr>
<th>pmol/L</th>
<th>n</th>
<th>Mean</th>
<th>min-max</th>
</tr>
</thead>
<tbody>
<tr>
<td>DA</td>
<td>3</td>
<td>544</td>
<td>292-777</td>
</tr>
<tr>
<td>TD</td>
<td>4</td>
<td>98.8</td>
<td>72.4-140</td>
</tr>
</tbody>
</table>

**Conclusions**

These preliminary data of normal values on venous umbilical cord blood appear to be useful for etiological issue and the management of DSD at birth.

**References**

Plotton et al. Horm Research, 2009
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