NEONATAL HYPO-KETOTIC HYPOGLYCEMIA SECONDARY TO TRANSIENT HYPERINSULINISM. DIAZOXIDE RESPONSIVENESS AND EXPERIENCE WITH FASTING TEST AFTER TREATMENT WITHDRAWAL

L. Salamanca Fresno*; N. Itza Martin*; C. Mora Palma*; J. Domínguez Riscart*; M. Sáez de Pipaón**; A. Campos Barros***; I. González Casado*. *Pediatric Endocrinology.** Neonatology Unit.***Institute of Medical and Molecular Genetics (INGEMM), IdiPAZ, CIBERER U753, ISCII. La Paz University Hospital. Madrid. SPAIN











Introduction:

- Transient hyperinsulinism is described in neonates with stress factors (intrauterine growth restriction (IUGR), large for gestational age (LGA), perinatal asphyxia, infants of diabetic mother etc.).
- Recognition and early treatment is prioritary to avoid neurological morbidity related with recurrent hypoglycemia.

58TH MEETING 19-21 SEPTEMBER 2019

Objetives:

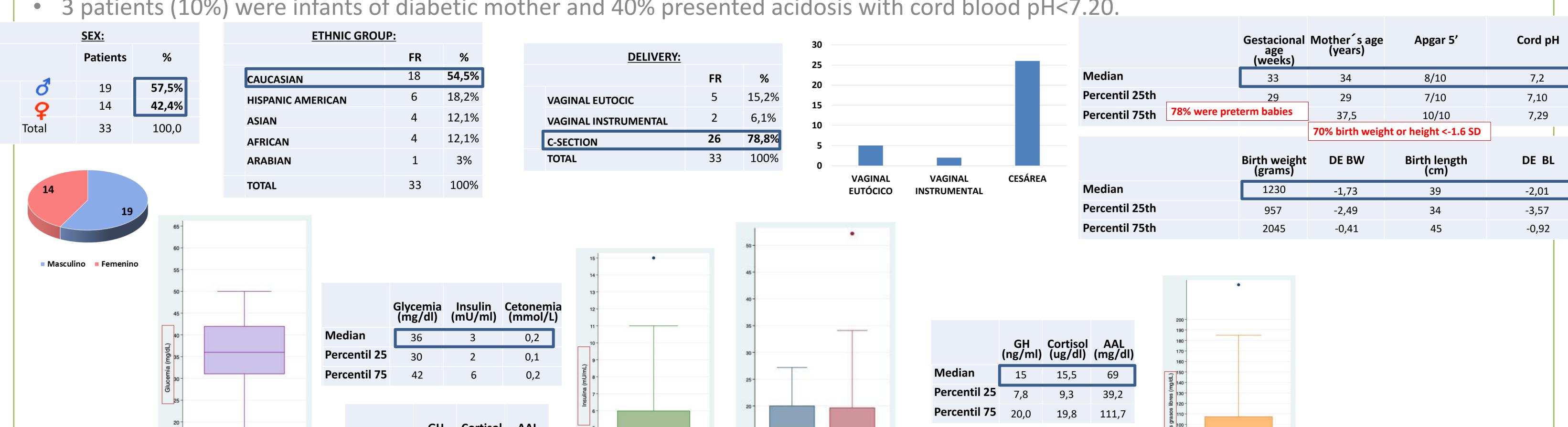
- Describe the incidence of transient hyperinsulinism.
- Clinical characterization and treatment response in neonates with hypoglycemia due to hyperinsulinism (monogenic forms excluded) admitted to a tertiary hospital NICU from January 2015 to April 2019.

Materials

- Prospective cohort study.
- Newborns > 7 days of age, with diagnostic criteria for hyperinsulinism: non ketotic hypoglycemia with detectable insulin levels, reduced free fatty acids, glucose infusion rate > 10mg/kg/min, and positive response to glucagon test).

Results (1):

- 3918 patients admitted, 0.76% (N = 33) presented hypoglycemia secondary to transient hyperinsulinism.
- The median diagnosis age was 23 days (IQR 10-29 days), with enteral feeding exclusively.
- 3 patients (10%) were infants of diabetic mother and 40% presented acidosis with cord blood pH<7.20.



- 87,9% received diazoxide treatment (dose ranged between 5-10mg/kg/day), presenting as most prevalent side effects hypertrichosis (81,8%) and edema (18,2%).
- Diazoxide median treatment duration was 83 days (IQE 41-110). The response was positive in 100%, with a fasting test performed on an outpatient basis, with glycemia > 60mg / dl after 10 hours of fasting after treatment withdrawal.
- Comparing preterm with term neonates, no significant differences were found regarding diazoxide treatment duration, maximum carbohydrate intake or resolution age.
- Molecular study was carried out through guided NGS in 70% of patients. No mutations were found so far in genes involving monogenic hyperinsulinism (ABCC8, KCNJ11, HNF4A, GLUD1, HADH, SLC16A1, GCK, UCP2, HNF1A, AKT2, INSR, CACNA1D), however, probably pathogenic variants were found in other candidate genes: G6PC2, TH, PMM2 and APPL1.

Conclusions:

- Transient hyperinsulinism is a prevalent entity to be considered in neonates with risk factors.
- In our series, term newborns presented transient hyperinsulinism (21% of patients) and newborns with weight and/or height appropriate for gestational age (28%).
- Low dose diazoxide treatment is effective. The fasting test could be useful for safe treatment withdrawal when resolution is suspected.



Poster presented at:







Median

Percentil 25

Percentil 75

20,0

19,8