Follow up results of a cohort of pregnant woman and their children with Inborn errors of metabolism backround.

Authors: Gutierrez, S., Hamui, E., Arbio, S., Andres, S., Di Meola, E., Martinez, C., Garcia, D., Baumberger, V., Astudillo A., Cannizzaro C.

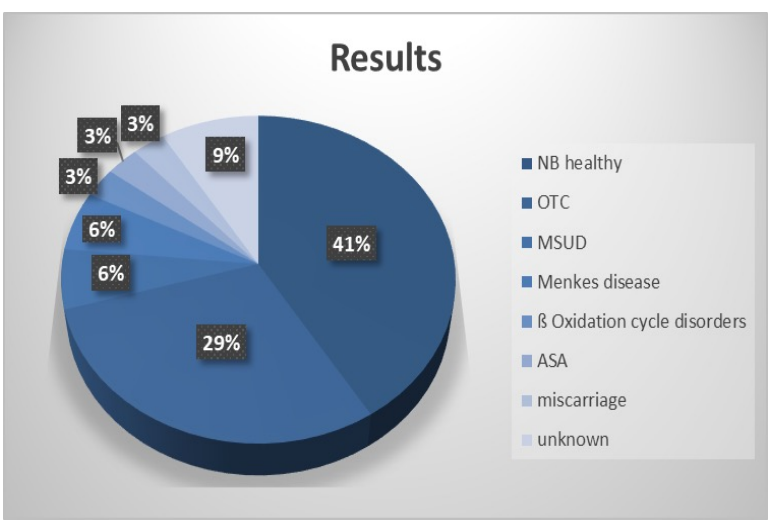
1. Hospital de Pediatría J.P.Garrahan. 2.Hospital Ramos Mejia. 3. Hospital Argerich.

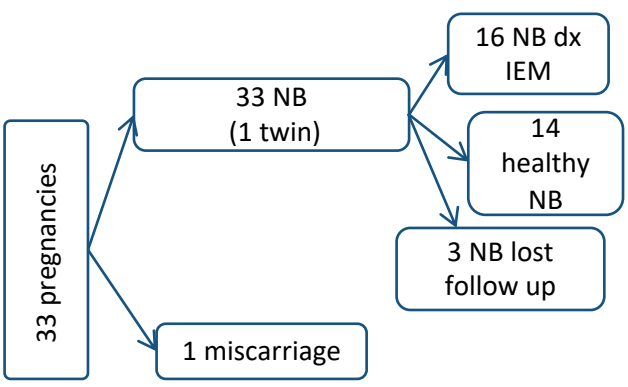
Introduction:
Inborn Errors of Metabolism (IEM) are "rare diseases" with a collective prevalence > 1:1800 newborns (NB). They are genetic disorders that involve a protein and/or enzyme of the metabolic process and can affect the mother-baby pair in a variable way. Pregnancy, and especially peripartum, are risky situations for pregnant women and affected newborns, therefore a multidisciplinary follow-up of the pairing is required to reduce morbidity and mortality.

Objective: To report the experience of monitoring pregnancies with personal or familiar history of Inborn Errors of Metabolism assisted in the "Fetal Diagnosis and Treatment Program" of a high-complexity pediatric hospital.

Methods: A descriptive and retrospective cohort study. Data collected from the medical records of pregnant women and newborns between 2010 - 2023.

Results: 33 pregnancies were followed in 31 pregnant women and 33 newborn (1 twin and 1 miscarriage). In 93.5% of the cases it was the 1st pregnancy. The Σ of Gestational Age (GA) at the consultation was 28 weeks (7 - 36.2), the Σ of maternal age 26 years (16 - 42).
25.8% of the pregnant women had a diagnosis of IEM: 6 cases (75%) Ornithine Transcarbamylase Deficiency (OTC), 1 case of Mucopolysaccharidosis Type I (MPTI) and 1 Maple Syrup Disease (MSUD). In the remaining 23 (74.2%), there was a family history of IEM, of these, OTC was more prevalent: 6 cases (26.1%) MSUD, Propionic Acidemia and Citrullinemia in 3 cases (13%) each one respectively, ß Oxidation cycle disorders 2 cases (9%) and Menkes Disease (MS), Glutaric acidemia, Congenital Methylmalonic Acidemia, Argininosuccinate Lyase Deficiency (ASA), Cobalamin Deficiency and MPTI in 1 case (4.3%), each. The pregnant women underwent dietary and metabolic care. Given the risk of hyperammonemia, prophylactic pharmacological treatment was added in the peripartum. There were no complications.
Pregnancies results: a spontaneous abortion and, of the live newborns, there were 2 twins (36 weeks of GA); the rest (94%) were born at term. In 16 newborns (48.5%) IEM was diagnosed: 10 cases of OTC, 2 with MSUD, 2 with MS, 1 with ß Oxidation cycle disorders and 1 with ASA. In 14 newborns (42.4%), IEM was ruled out and in 3 newborns (9%) the diagnosis could not be completed. Of the total number of newborns affected, there were 3 deaths (9%) and in 3 cases follow-up was lost.





Σ	Age
GA	28 weeks (7-36.2)
Maternal age	26 years (16-42)

Conclusion
In this cohort, OTC deficiency was the most frequent condition, both in diagnosed pregnant women and in those with only a family history of this disease. Almost half of the newborn presented some Inborn Errors of Metabolism (IEM), constituting twice as many cases as in diagnosed pregnant women. OTC deficiency was also the most frequent condition. The survival of children with IEM is increasing, reaching childbearing age with a good quality of life. Information on IEM and pregnancy is very limited. Work should be done on the investigation and prevention of metabolic complications in the mother-son binomial, in multidisciplinary teams with the participation of experts.