INTRODUCTION

- In North-Eastern Italy newborn screening for congenital adrenal hyperplasia (CAH) has been performed since 2001.
- The screening program based on 17-OHP levels in dried blood spots has made early diagnosis for classic form of 21-hydroxylase deficiency (21-OHD) possible, allowing precocious treatment and reduction of mortality rate.
- A high false positive rate at screening test, especially in preterm, low-birth-weight and critically ill newborns, is described.

AIMS OF THE STUDY

- To summarize the results of the newborn screening for CAH held in the last 18 years in North-Eastern Italy.
- To evaluate the diagnostic utility of simultaneous determination of 17-OHP, cortisol, 11-deoxycortisol, delta 4-androstenedione and 21-deoxycortisol by liquid chromatography-tandem mass spectrometry (LC-MS/MS) as second tiers test performed on the same blood spot.

METHOD AND MATERIALS

- Since 2001 dried blood spots from newborns have been screened with a time-resolved fluoroimmunoassay method (DELFIA) for 17-OHP determination.
- Over the years, the cut-off levels of 17OHP have been adjusted according to gestational age.
- Since 2017, samples resulted above the cut-off have been immediately analyzed by LC-MS/MS in order to differentiate affected patients from false positive newborns.

RESULTS

- Since 2001, 786,302 newborns have been screened.
- 34 diagnosis of classic form of 21-OHD and a total incidence of 1:23126.
- The subsequent adjustments of cut-off values for 17-OHP based on gestational age and the use of LC-MS/MS, as a second-tier test for positive CAH screening, have significantly reduced the recall rate.

CONCLUSIONS

- Screening for CAH proved to be useful in the neonatal diagnosis of classic form of 21-OHD, allowing a precocious treatment and significantly reducing mortality.
- The use of LC-MS/MS as a second tier test improved the positive predictive value of the screening program.
- LC-MS/MS cannot be seen as a replacement for the conventional method, but it is extremely useful as a second-tier test, in particular in preterm, low-birth-weight and critically ill neonates, preventing unnecessary blood draws, medical evaluations and stress to families.