

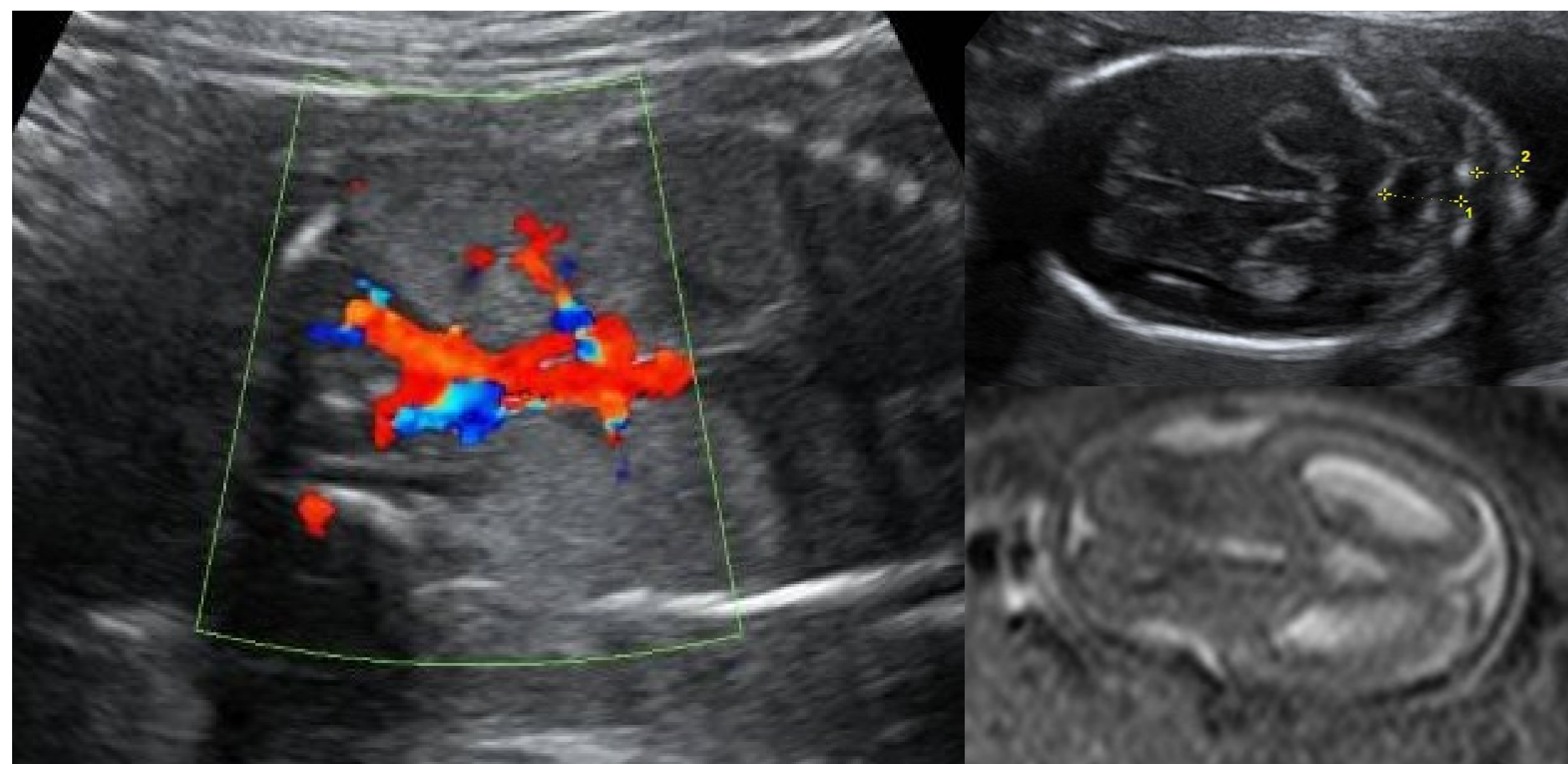
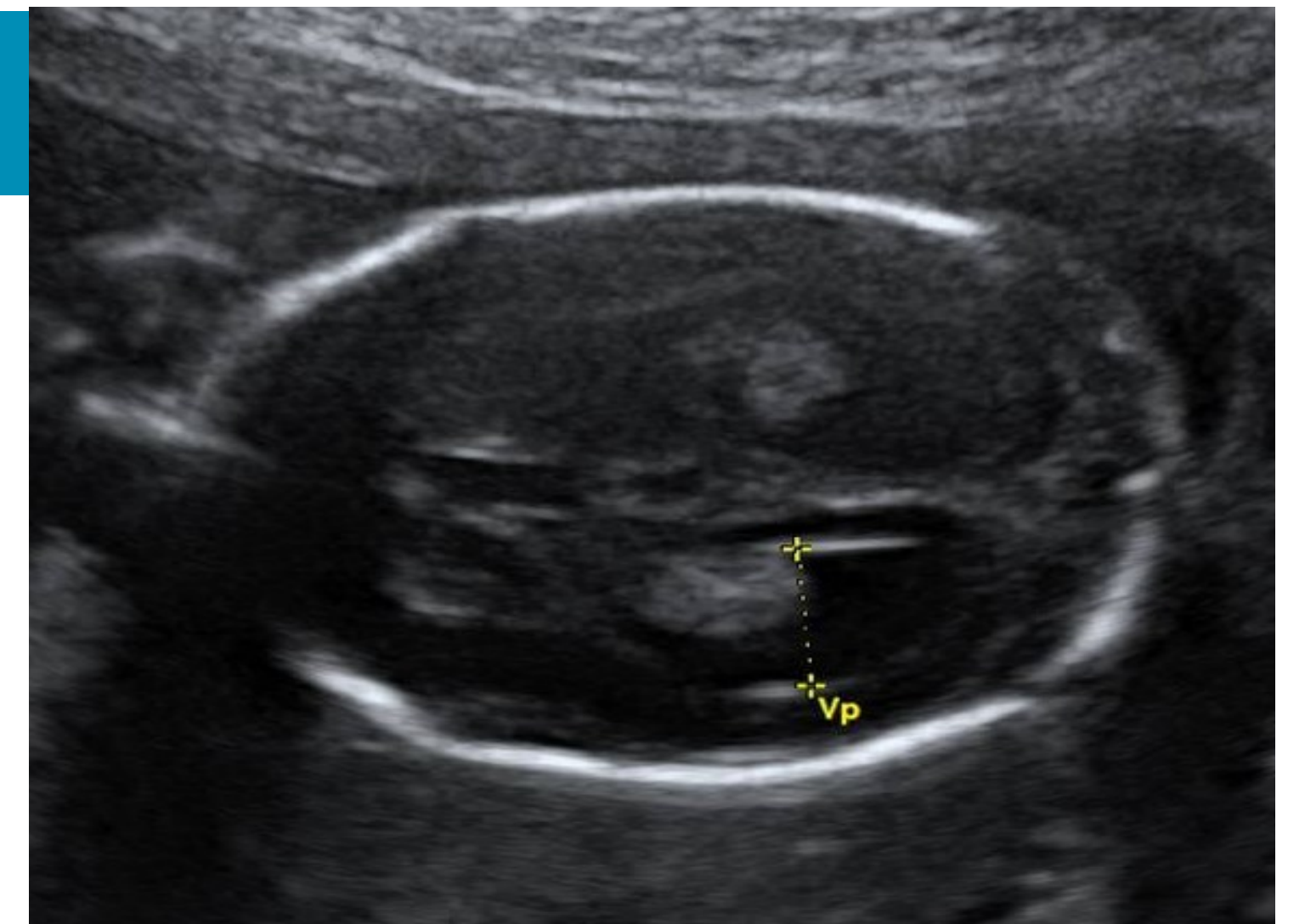
## PRENATAL DIAGNOSIS OF CARNITINE PALMITOYLTRANSFERASE II DEFICIENCY

RAMIREZ PUJADAS A, BAGAN ROBLEDO H, SOULLIMAN N, GUILLEMAT FONT Y, ARAGON CASALET P, GIL ROMERO E  
Departments of Obstetrics and Gynaecology  
Hospital General de Catalunya - IDC Salud; Spain

### BRIEF INTRODUCTION

Carnitine Palmitoyltransferase II Deficiency due to mutations in the CPTII gene is often diagnosed in postnatal period after more than one terminated pregnancies for polymalformation and in successive stillbirth or perinatal loss cases.

It can happen in adolescence, infancy and neonatal period. The neonatal presentation includes several cerebral and renal malformations and it is often fatal in the first days of life.



### OBJECTIVE

- To demonstrate the importance of
  - prenatal diagnosis to ensure a correct assessment.
  - the advance in new molecular genetic techniques in detection of rare congenital diseases.
- To perform prenatal differential diagnosis of a bilateral multicystic kidneys

### CLINICAL CASE

A 38-year-old woman, gravida 2 para 1, low risk pregnancy.

- Ultrasound examination at 20 weeks showed **enlarged hiperechogenic kidneys, ventriculomegaly, mega cisterna magna and oligohydramnios.**
- Fetal RM confirmed the alterations.
- Chromosomal microarray analysis was normal.

The pregnancy was terminated at 22.4 weeks' gestation.

- Necropsy confirmed ultrasound findings.

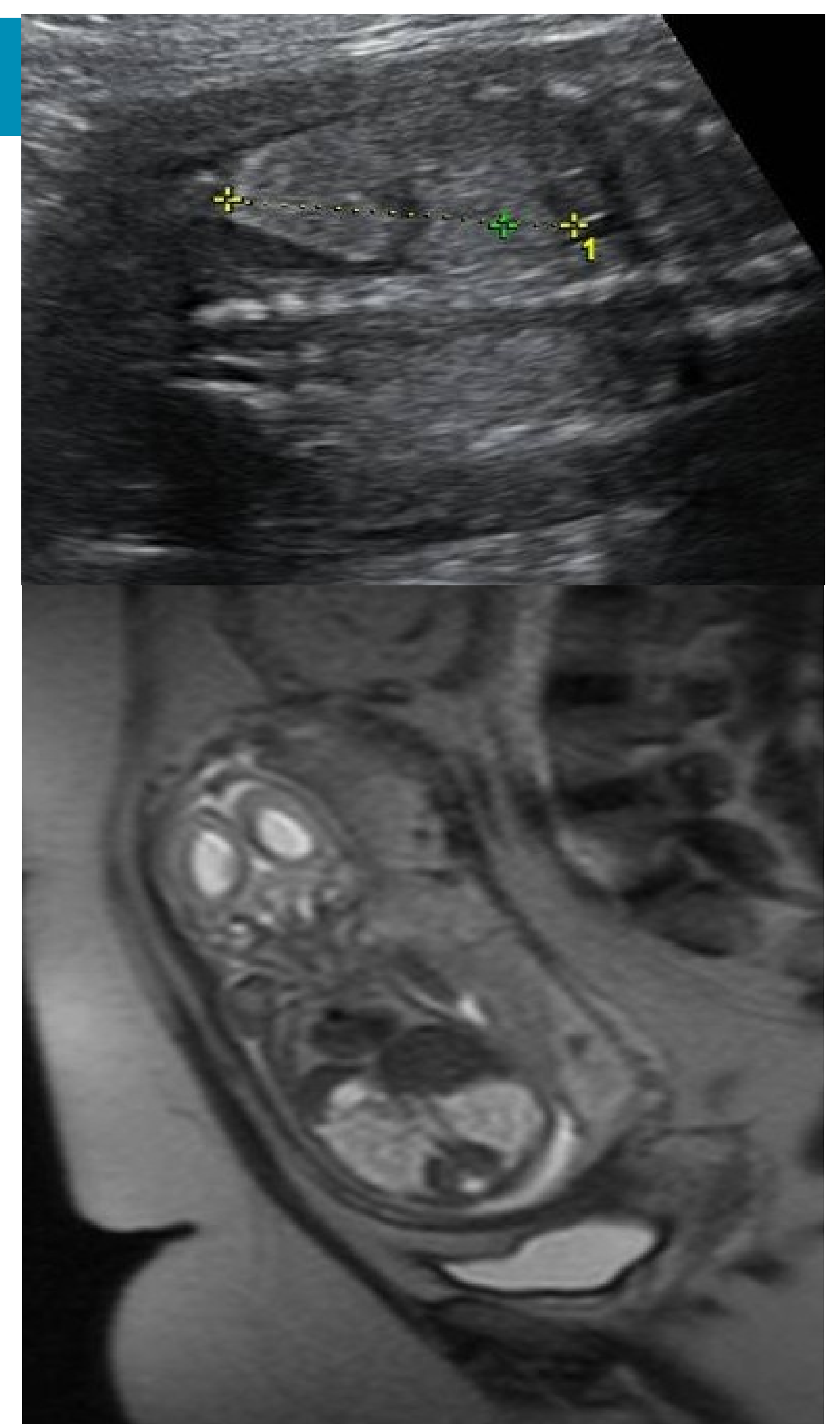
Postnatal PKHD1 and 140 targeted renal gene panel study were also normal.

She spontaneously became pregnant two years later. She underwent chorionic villus sampling at 11 weeks with no result because of sample contamination. Normal scan at 13.5 weeks was performed.

- Ultrasound examination at week 18.1 diagnosed the same malformations as the previous pregnancy.

The pregnancy was terminated at 18.5 weeks.

- Whole exome sequencing diagnosed CPTII deficiency.



### CONCLUSIONS

- CPT2 deficiency may be underestimated and should be considered when ultrasound examination shows antenatal brain and kidney malformations.
- Molecular diagnosis in suspected cases should be done in order to improve prenatal management.
- Looking for congenital disorders by genetic study and an exhaustive ultrasound must be conducted to search other malformations and to assess future pregnancies.