Objective
Exome sequencing is a powerful tool to identify disease causing mutations in fetuses with malformations and unrevealing molecular karyotype. We applied exome sequencing in a fetus with hydrops, congenital heart disease and gonadoblastoid testicular dysplasia.

Methods
Whole exome sequencing of a fetal sample was conducted on a HiSeq 2000. Confirmation analysis was performed by using conventional Sanger sequencing.

Results
Whole exome sequencing revealed a pathogenic mutation in the RIT1 gene (c. 270G>A (p. Met90Ile)), leading to the diagnosis of Noonan syndrome type 8. Comparison with the parents revealed that the mutation occurred de novo in the fetus.

Conclusion
Here we present the first case of a fetus with RIT1 mutation leading to fetal hydrops and lethal outcome. Furthermore, the combination of heart malformation and fetal hydrops together with gonadoblastoid testicular dysplasia may represent a distinct subtype of Noonan syndrome. This case demonstrates the power of exome sequencing to identify disease causing mutations in unclear cases and to establish the recurrence risk for further pregnancies.