A case of TAR syndrome (trombocytopenia with absent radius syndrome)

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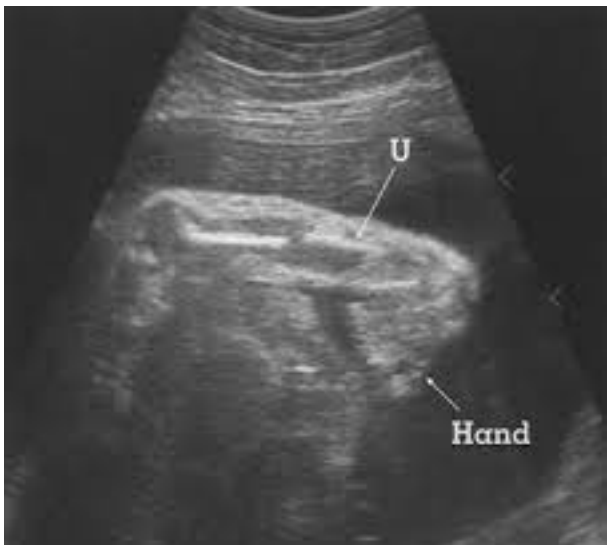
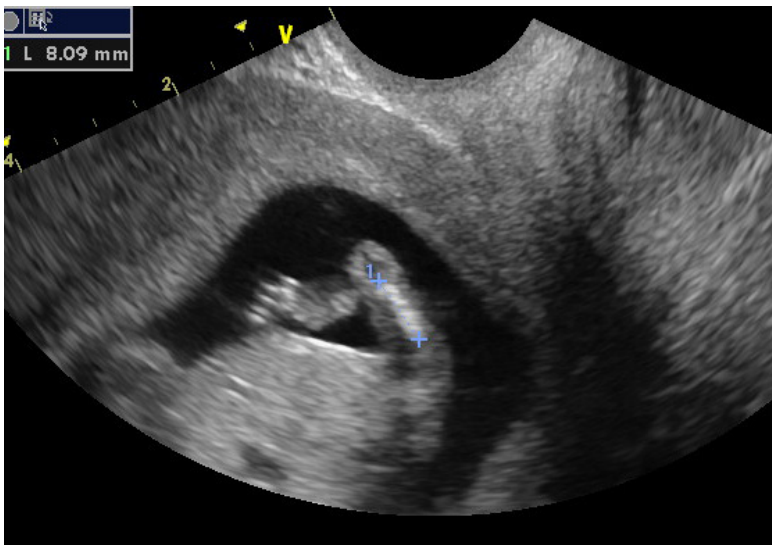
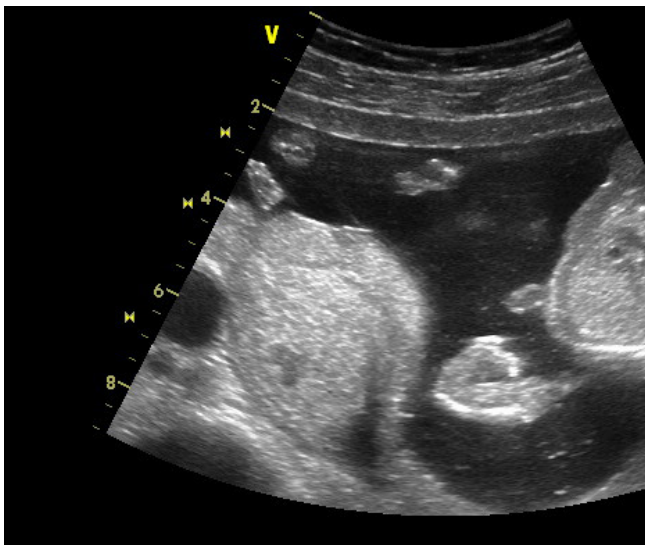
TAR syndrome is a rare genetic disorder that is characterized by the absence of the radius bone and dramatically reduced platelet count. The radii of both arms are affected bilateral. A variety of sceletal abnormalities occur in individuals with TAR syndrome. This syndrome may occur as a part of the 1q21.1 deletion syndrome. The incidence of TAR syndrome is fever than 1 in 100,000 newborns.

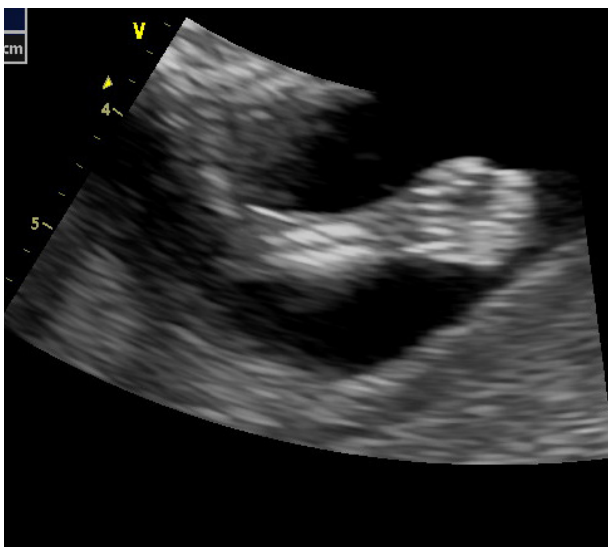


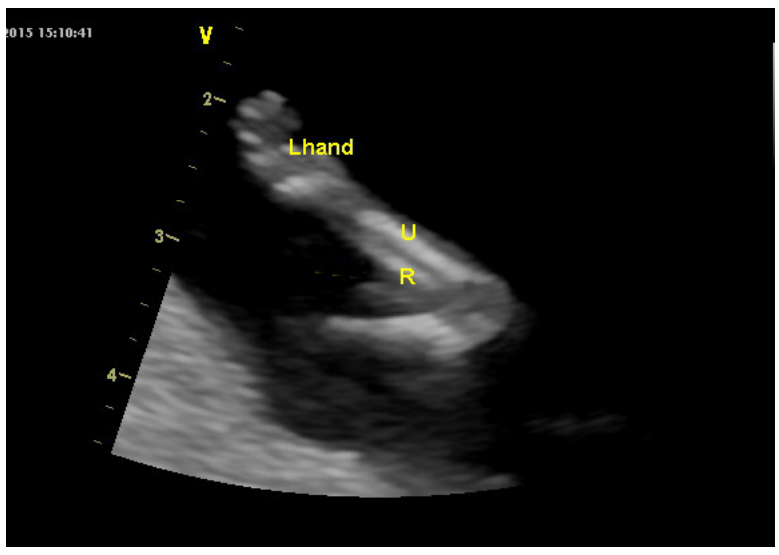


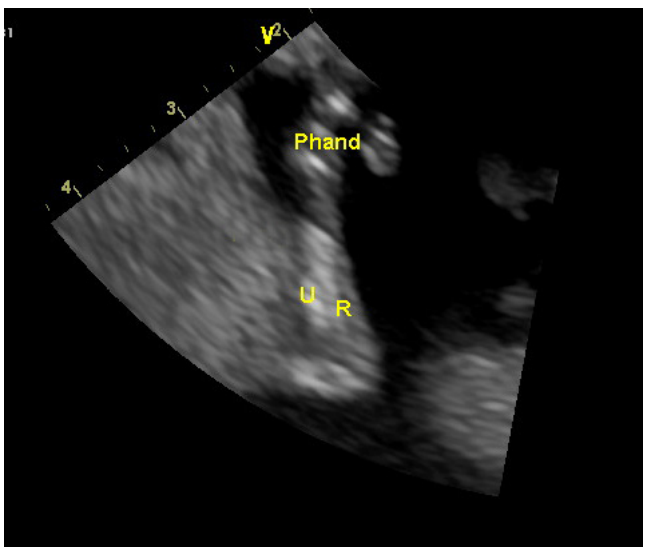


Genetics
TAR syndrome is inherited as an autosomal recessive genetic disorder and caused by two different type of mutations of chromosome 1, in the RBM8A gene.
A 2007 research article identified a region of chromosome 1, 1q21.1, containing 11 genes including besides RBM8A.
One of the methods of an early prenatal diagnosis, which has been applied in this case, is the preimplantation genetic diagnosis (further PGD). Principle of PGD- the couples with a carrier of a chromosomal aberration (TAR,…) will undergo the IVF cycle. On the 3th day after the fertilisation, when the embryos achieve a stage of eigth cells, the embryologist collects 1-2 cells from every embryo. These cells undergo next steps of the genetic examination. The FISH (fluorescent in situ hybridization) is a more sensitive cytogenetic method. It is based on the principle of hybridization of a short fluorescently labelled sequence of DNA (a so-called probe) with the corresponding sections of the tanged DNA sequence from the embryonic cells being examined.

A case report
29 year old women, 4th gravida 0 para with a history of two premature terminations of pregnancy for a diagnosis of TAR syndrome. The women came at our centre before the 4th planned pregnancy. She was included in the IVF programme with PGD.
From the obstetric anamnesis- 1st pregnancy (2010)- terminated in 22nd week of gestation for ultrasound diagnosis of the absence of the radius and the deformation of forearms, from the fetal DNA was detected deletion 1q21.1, the same microdeletion was proved from the patient´s (mother´s) DNA, from DNA of her husband wasn´t detected this deletion. 2nd pregnancy (2012)- missed Ab in 9th week. 3rd pregnancy (2013)- terminated in 14th week for US diagnosis of TAR syndrome. 4th pregnancy (2015)- 1st time in Gennet, the women was included in the IVF programme, this pregnancy was from 2nd cycle of IVF with PGD (1st cycle of IVF – any embryo for a embryotransfer), 1st trimester with normal sonoanatomy and biometry, CRL 58mm, BPD 19,5mm, NT 1,3mm, normal biochemic screening, the next control- dg missed Ab in 16th week- fetus with the normal sonoanatomy and normal karyotype from DNA testing, a deletion 1q wasn´t proved by haplotype analysis.







In conclusion
TAR syndrome belongs to an autosomal reccesive genetic disorder with chromosomal abnormality. The first step to a succesfull pregnancy is molecular testing of the deletion/duplication analysis for the region of the chromosome band 1q21.1 that contains RBM8A gene. By use PGD it is possible to reduce the risk of spontaneous abortions by means of a selection and transfer of the embryos, which are not affected by any severe chromosomal aberration or another genetic disease. Unfortunataly, a my case wasn´t with the happy ending, but maybe… next the parents may start the pregnancy with a feeling that their future descendant will not suffer from any several genetic condition or a defect occuring in the family, which can be examinated by the methods of prenatal diagnostics.