

Is it always possible to determine a diagnosis? Prenatal ultrasonography, post mortem magnetic resonance, autopsy

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Abstract

Pathological-anatomical autopsy is the gold standard for determining of foetal abnormalities, but in some cases its role is limited (pathology of central nervous system, in particular, in case of ventricular dilatation or developed autolysis). In pathology of central nervous system, where insufficiency of autopsy can occur, additional post mortem magnetic resonance imaging (MRI) is performed to determine type of malformation.

In this case report, we would like to point out the fact that although all investigating methods including post mortem magnetic resonance and autopsy (incl. imunohistochemical tests) are used, this need not necessarily result in a clear diagnostic conclusion of the aborted foetus. Post mortem MRI visualized pathology: dilatation of both lateral ventricles, more in the left and, above all, a pathological focus parasagittally on the right with haemorrhage and cystic component; it raised a suspicion on ependymoma. However imunohistochemical test did not give an unambiguous conclusion; therefore diagnosis based on MRI could not be uniquely verified.

CASE REPORT

27 years old primipara, present pregnancy without complication. Sonographic examination of foetal morphology at the end of the 1st trimester within first-trimester screening of congenital defects and

chromosomal aneuploidies was negative. Sonographic examination in the 20th week described asymmetric dilatation of the right lateral ventricle and dilatation of the 3rd ventricle (Figure 1). The

pregnant woman was informed of the findings and she asked to terminate the pregnancy in accordance with the valid legislation. Abortion was induced in the 22nd week, post mortem MR was performed immediately and was followed by autopsy.

The MR examination was performed in all orthogonal planes in a T2W image with the following parameters: TE 120 ms, TR 5272 ms, thickness of slice was 3mm (THK (3.0/0.3 mm)). Post mortem MR showed supratentorial focus parasagittally on the right, according to the MR signal haemorrhagic and most probably cystic character of the focus (post mortem MR is performed routinely only in T2W image, so that it is not possible to determine exactly the character of the substrate) (Figure 2 A,B). Subarachnoid bleeding was also observed, lateral ventricles were dilatated, mostly on the left side, the 3rd ventricle was of normal width. Post mortem MR described the focus mentioned above as atypical, possibly a cyst (arachnoid or glioepithelial), we were not able to explain the bleeding in this diagnosis or in the differential diagnosis, we considered a tumour with cystic component and presence of bleeding.

The autopsy demonstrated haemorrhage in cavum septi pellucidi, dilatation of lateral ventricles and in the right lateral ventricle a fragile rosy focus 8×10×8 mm was found, which was connected with germinal matrix. It was placed closely under ependymoma and was formed from small uniform cells in the histological examination. Most probably it could be medulloblastoma, cellular ependymoma or hyperplasia of the germinal matrix. An immunohistochemical test of this partially autolysed material did not confirm any of these diagnoses unambiguously. For this reason the autopsy could not provide a clear conclusion about the focus in the right lateral ventricle. The genetic test was negative, normal karyotype (XY) was determined by an examination of the amniotic fluid.



Fig. 1. Prenatal ultrasonography, 20th week, described asymmetric dilatation of the right lateral ventricle and the 3rd ventricle.

DISCUSSION

Unfortunately none of the examinations provide us with clear information about the pathological focus. Prenatal ultrasonography described asymmetric dilatation of the right lateral ventricle, the pathological focus was incorrectly interpreted as a dilatation of the 3rd ventricle. Magnetic resonance showed this pathological focus parasagittally on the right, the 3rd ventricle was not dilatated and this was also confirmed by autopsy. MR displayed asymmetrical dilatation of both lateral ventricles, more in the left, possibly caused by the presence of the pressure of pathological focus on the paths of the cerebrospinal fluid. In order to be able to compare results of autopsy and post mortem MR, routinely we do not inform about the results of MR before performing autopsy, and after finishing both methods we compare and determine whether the conclusions are identical and which method brings more information (Griffiths *et al.* 2005; Whitby *et al.* 2006). Autopsy determined dilatation of lateral ventricles with bleeding into cavum septi pellucidi and pathological focus in the right lateral ventricle, which was closely under the ependyma and was formed from small uniform cells. This could concern ependymoma, medulloblastoma or hyperplasia of the germinal matrix. With regard to the fact that immunohistochemical examinations of necrotic material are sometimes insufficient, it was not possible to specify the specific pathological unit. From the point of view of MR imaging, the finding did not correspond to typical picture of medulloblastoma (atypical localisation of bleeding, cystic component) (Osborn *et al.* 2004). Localisation of the focus indicated ependymoma, there may be presence of bleeding, cysts, though this is very rare intrauterinally (Garel 2004; Osborn *et al.* 2004; Rickert *et al.* 2002). Localisation of bleeding testifies to uncomplicated bleeding into the germinal matrix, against this there was a cystic component of the focus. Autopsy and post mortem MR are performed to provide us with final information about the type of congenital malformation or in this case about the type of cerebral tumor to give parents relevant information about the risk of repeating this malformation in future pregnancy, a second no less important reason is to specify prenatal ultrasonographic diagnostics on the basis of the feedback (Griffiths *et al.* 2005; Sebire 2006; Vaneckova *et al.* 2010; Whitby *et al.* 2006).

This case report points out that although all investigating methods including post mortem magnetic resonance and autopsy are used immediately after abortion, this need not necessarily result in a clear diagnostic conclusion.

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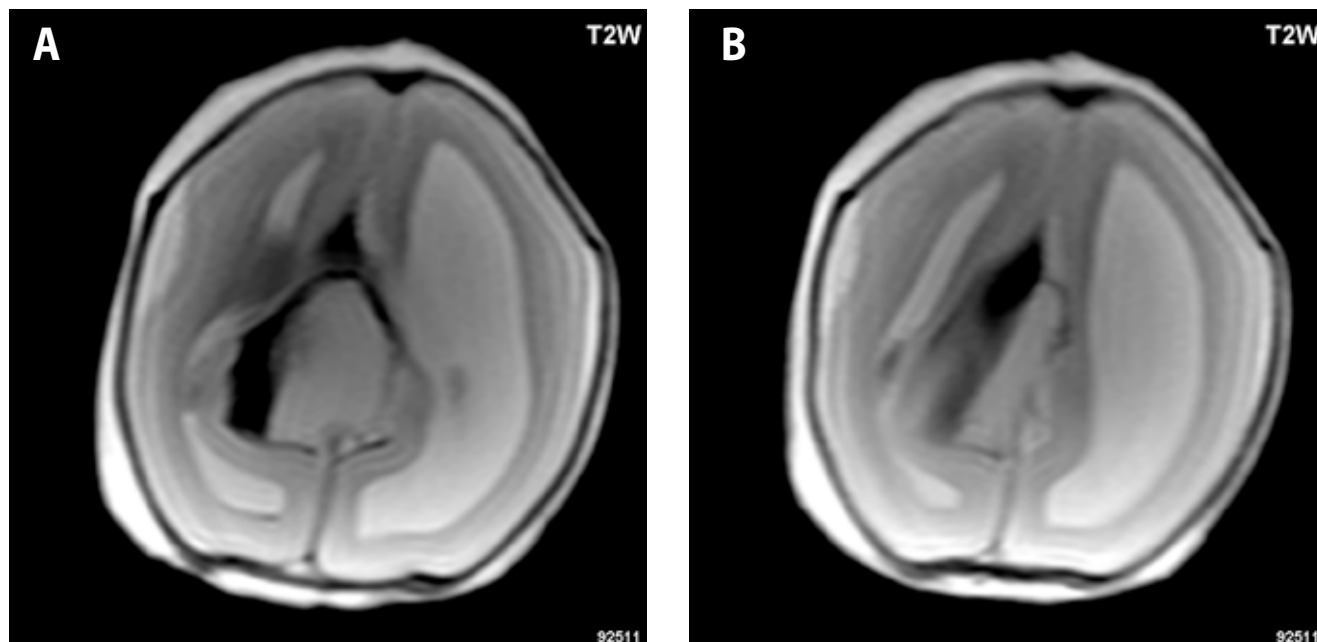


Fig. 2 A,B. Post mortem MR, supratentorial focus is evident parasagittally on the right, according to MR signal with bleeding, character of the focus is most probably cystic. There is subarachnoid haemorrhage as well, the lateral ventricles are dilatated, more on the left, the 3rd ventricle is of normal width.

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