SHOULD GENETIC PRENATAL DIAGNOSIS BE PREFORMED?

CLINICAL CASE

Manuela, a 38-year-old woman with a history of breast cancer, is in her 16th week of pregnancy. Various forms of breast cancer exist in her family. She and her husband are monitoring her pregnancy through a public hospital and if the sex of the fetus is female, they would like to have a prenatal genetic screening test to rule out the possibility that it has the genetic mutations BRCA1 and BRCA2. If the fetus has these mutations, they would like to terminate the pregnancy. The hospital's gynecologist took this case to the Ethics Committee with the following question: Should we perform this prenatal genetic screening test at our hospital?

ETHICAL ANALYSIS

The primary values in conflicto, in this case, are: on the one hand to respect the parent's wish to perform the prenatal genetic testing (PGT) and on the other hand, the adequate distribution of financial resources, because PGT is very expensive, and also the protection of life in utero. In order to perform the PGT it would be necessary to practice an amniocentesis, which is not without risks and as a result, can lead to complications such as an abortion.

In this case, the first thing to clarify is that being a carrier of BRCA1 and BRCA2 does not meet the criteria for a legal voluntary termination of pregnancy (VTP). Being a carrier of BRCA1 and BRCA2 is a risk factor for having an adult disease (gynecological cancer), however, it is not a disease. The mutations of BRCA1 and BRCA2 do not necessarily imply that their daughter will develop this disease, rather, that she is at risk to be predisposed to the appearance of some cancerous pathologies, mainly breast and ovarian, but not all patients with the mutation will suffer from breast and ovarian cancer. It would not be the same, if for example, the fetus had Thalassemia Major, or Trisomy 21.

POSSIBLE COURSES OF ACTION

- Not perform the prenatal genetic screening test; as being a carrier of BRCA1 and BRCA2, does not meet the criteria to voluntarily terminate the pregnancy (VTP).
- Study if the mother meets the risk criteria in order to carry out a genetic screening test for mutations BRCA1 and BRCA2. If so, perform the genetic screening test on the mother.
- If the mother is a carrier of the mutation, perform the prenatal genetic screening on the fetus. If she is not a carrier, then refuse to perform the test on the fetus.
- Inform the patient and her husband about the implications of the mutations BRCA1 and BRCA2 in detail, then verify that they have understood the information.
- If the fetus is a carrier of the mutation, do not carry out the termination of the pregnancy.
- If the fetus is a carrier of the mutation, perform the abortion.

RECOMMENDED COURSES OF ACTION

- Inform the patient and her husband in detail about the implications of a mutation in BRCA1 and BRCA2. Inform them of the criteria for a VTP and verify that they have understood the information.
- The prenatal Genetic screening test cannot be performed: being a carrier of BRCA1 and BRCA2 does not meet the legal criteria to voluntarily terminate a pregnancy (VTP).
- Furthermore, with the current regulations in Spain it wouldn't be possible to have an abortion as her pregnancy has passed the legal limit for voluntary abortion (up to 14 weeks) and the presence of a mutation that increases the risk of an illness is not justification to voluntarily terminate a pregnancy due to medical reasons.
- If the mother meets the risk criteria, the genetic screening test would be performed on her, and if she is a carrier, the recommendation would be to perform the genetic screening on her daughter after her birth.

DISCUSSION

In the case being studied, a VTP could not be authorized for being a carrier of BRCA1 and BRCA2. Putting a fetus at risk without any real consequences (a VTP could not be performed) would not be acceptable in this case, and performing an amniocentesis would not be indicated. Here, a question arises: What if it were in the 10th week of pregnancy? In the public health system the PGT would not be authorized, since being a carrier of BRCA1 and BRCA2 does not meet the criteria for a VTP. In a private center, perhaps the test could be performed (first it would be performed on the mother) and then depending on the results, the parents could choose to have a VTP, since they would be within the legal limit (less than 14 weeks).

Therefore in the case that is suggested above, it is essential that the patient and her husband first have all the necessary information regarding the test and about the criteria for a VTP. It is also important that they know and understand what procedures are necessary to perform the PGT, the possible complications, and the usefulness of the results at the present time.

Regarding whether the test should be carried out on the mother to see if she is a carrier of the mutation BRCA1 and BRCA2, it can be done, however, in this case there would be no subsequent consequences to the fetus since, regarding the results, neither a PGT nor a VTP could be performed. If the mother meets the criteria for the risk, then she could have the genetic testing done, and if the result of that test is positive, then after the birth of her daughter the test may be performed on the baby following the current guidelines. In the event that the patient's daughter was a carrier of the mutation, early screening protocols should be applied.

This case has been studied in accordance with the current legislation in Spain, therefore its resolution may vary in other regulatory frameworks.

Sgd.: ASISA-Lavinia Bioethics and Health Law Committee

28th of April, 2023