

2016

Case No. 26 - APLS

New York Law School

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Recommended Citation

New York Law School, "Case No. 26 - APLS" (2016). *Patient Safety Project*. Book 27.
http://digitalcommons.nyls.edu/patient_safety_project/27

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APLS

37 year old G1 P0 who presented to her general ob/gyn for an anatomy scan. She had previously refused amnio or genetic testing for advanced maternal age. At the time of the ultrasound, borderline large lateral ventricles were noted in the brain. The technician also said that the fetus looked like a female and suggested that she get a consult with the local MFMs. The local MFM did the consult, thought the anatomy looked fine but the parents at that point opted for an amniocentesis to put their minds at rest. Again the technician thought the fetus was likely female. The amniocentesis was done and the FISH (fluorescent in-situ hybridization) came back in two days saying normal male. The patient was called by the MFM's genetic counselor and was relieved however when she spoke to her relative, also a physician, he cautioned that it was unusual that two ultrasounds said female and the result of the FISH upon which she was counseled not to make any permanent decisions without waiting for the full report of the chromosomes was male. She called the genetic counselor back who said that ultrasound determination of sex could be wrong but advised that she return immediately for another ultrasound. The MFM said that the genitalia still looked to him like a female but advised waiting for the full karyotype and added some additional tests saying it could be a microphallus or bifid scrotum. He asked the genetic counselor to call the lab and the lab again sent a fax with the normal male karyotype from the FISH.

The patient and her husband spoke to the genetic counselor and learned that FISH is unlikely to be wrong and what some other possible diagnoses were if there was discordant genitalia and chromosomes.

The patient who was approaching the legal limit of termination, decided to abort and returned to her general ob/gyn for a D&E without telling the MFM.

The patient was scheduled and had her D&E by the general ob/gyn. The full karyotype came back on the day the procedure was performed and the geneticist realized that the FISH report was a typo, which had been faxed twice.

The patient was devastated upon learning they had terminated a normal female.