Another kind of 'second opinion' for newly diagnosed cancer patient.

There is no underestimating the shock of receiving a life-changing diagnosis: obtaining a second opinion is always recommended. What might also be recommended is to confirm that the slide/sample is really from the patient; really another type of 'second opinion'.

As a forensic DNA testing laboratory (a real-world version of the television CSI just not as good looking), we are used to seeing quite a variety of samples – different types, different sources and different reasons for DNA testing. Recently we have been asked to use DNA-based identity testing in order to verify that a pathology sample, used in a diagnosis of cancer, was really from the patient. This testing does not address the diagnosis, only a second opinion from a pathologist can do that, but it does tell the patient (and the physician), that the tissue sample/biopsy really belongs to that patient.

Laboratory errors are an unfortunate fact of life: to err is human and there are several good studies that have measured the types and number of errors in pathology laboratories. The results are a little scary: up to 1% (one in a hundred) of pathology samples can have a mis-identification error (there are actually two kinds of identification errors). This may not be the easiest conversation to have with your oncologist, but they will certainly be aware of this issue. Happily, there is a good solution to the problem of sample mis-identification.

Using DNA analysis, just like forensics or paternity testing, a modern DNA lab can compare the DNA profiles of the pathology sample with the patient's own profile. If they match then that is the correct sample –if they don't then someone has to find the correct biopsy. This kind of verification is reasonably priced (in the \$250 range), might be covered by insurance and is fast (results in 2-3 days at the most). Given the possibility of a life-changing diagnosis, sample verification may be something that you might consider looking into. Newly diagnosed cancer patients have enough on their plate – they can at least be sure that it is their sample that was examined. For those who want to delve into this subject in more detail, the following references are

a place to start.

References:

1) Rate of occult specimen provenance complications in routine clinical practice. Pfeifer, J. D. and Jingxia, L. (2013) American Journal of Clinical Pathology 139, 93-100.

2) The Changing Spectrum of DNA-Based Specimen Provenance Testing in Surgical **Pathology.** Pfeifer, J.D. et al., (2011) Am J Clin Pathol 135(1):132-138.

3) Mislabeling of cases, specimens, blocks, and slides: a College of American pathologists Study of 136 institutions. Nakhleh, R.E. et al., (2011) Arch Pathol Lab Med. 135(8): 969-74.

4) Specimen Labeling Errors in Surgical Pathology. An 18-Month Experience. Layfield, L.J. et al., (2010) Am J Clin Pathol 134(3): 466-470.

5) Tissue Floaters and Contaminants in the Histology Laboratory. Platt, E. et al., (2009) Arch Pathol Lab Med 133: 973-978.

6) Contamination of Histology Biopsy Specimen - a potential source of error for surgeons: a case report. Burke, N. G. et al., (2009) Cases Journal 2:7619.

7) Identifying Cross Contaminants and Specimen Mix-ups in Surgical Pathology. Hunt, J.L. (2008) Adv Anat Pathol. 15(4):211-217.

8) Identification of Biological Samples in a Case of Contamination of a Cytological Slide Preparation. Junge, A., et al., (2008) Journal of Forensic Science 53(3):739-741.

9) Diagnosis of Ectopic Tissue Versus Contamination by Genetic Fingerprinting in a Routine Surgical Pathology Specimen. Venditti, M., et al., (2007) Human Pathology 38(2):378-382.

10) Patient Identification Error Among Prostate Needle Core Biopsy Specimens – Are We Ready for a DNA Time-Out? Suba, E.J. et al., (2007) The Journal of Urology 178(4 Pt 1): 1245-1248.

11) Forensic DNA Typing: Biology, Technology, and Genetics of STR Markers. Butler, J.M. (2005) Elsevier Academic Press.

12) Development of a decision-analytic model for the application of STR-based provenance testing of transrectal prostate biopsy specimens. Pfeifer, J . D. et al., (2012) Value Health. 2012;15:860-867.