FAQS from Dr. Churpek's webinar

My spouse was diagnosed in 2014; his brother had a blood cancer and his father had a different organ cancer. We were advised to just have our adult children continue having their annual well checkups. Fast forward, they each were tested and both are donors for his stem cell transplant. Has more recent research indicated that the children should now get more genetic testing to see if they are carriers or risk? They are now age 32 and 37 and have no current symptoms.

In these cases, we evaluate the person with the blood cancer first (your spouse). If he and his brother both had MDS or AML, we would consider genetic testing for your spouse to help understand if there is a genetic component and then can best advise the rest of the family. It is very reasonable for your children to have annual visits and a blood count every year (this will not detect all blood cancers and is not of proven benefit in this situation, but can help find low blood counts that require more work-up in the familial setting).

How would I know if my MDS with tp53 mutation has a familial tendency?
My niece developed her 2 breast ca with BRCA1 and BRCA 2 mutations, had radiation, her Dad had Multiple Myeloma. Is she at risk for MDS and does she need genetic counseling?

TP53 mutations are relatively common in MDS and are most often acquired in the MDS (meaning they are usually only in the MDS cells and not in the rest of your body so not passed down to children or causative of other cancers). your niece has had inherited genetic testing (it sounds like) so she may have had TP53 on the genetic testing if she had a gene panel (a list of many genes tested all at once) which is commonly done now. Carrying both a BRCA1 AND BRCA2 mutation in all of one's body cells is rare but can happen. We would usually test her parents next to see which parent carries the BRCA1 or BRCA2 mutations and then offer testing for these to other at risk relatives (you could get your own testing for these genes as your children would then be at risk for carrying them). You should see a genetic counselor or cancer risk clinic to help sort this out.

How do you become a part of a familial MDS study?

You can contact my research assistant, Lauren Lovrien at lnmcnair@wisc.edu.

If a person has had MDS but there are no personal risk factors or family history patterns, can you give a percentage for the likelihood that the person's children could develop MDS?

The risk would be expected to be very small. If someone had MDS at a young age (ie. 50 or younger), this may make that small number higher but exact estimates for this situation not known.

Doesn't a single gene mutation need to work with other genes to be significant to health? Are the other genes more likely to be normal or abnormal?

This question is hard to answer because it is complex. Usually a single gene mutation works with others to contribute to cancer but in the inherited setting, a single gene mutation can be significant to health. For example, carrying one abnormal copy of BRCA1 increases a woman's lifetime breast cancer risk from

12% to between 30 and 87% and ovarian cancer from 1% to up to 27%. This is all from the effect of one single gene—again, not a guarantee that something will happen but a substantial change in risk that we act on to prevent cancer.

Your statistics showing 1 in 8 have a close relative with blood cancer. My mom died from Multiple Myeloma. Would it be helpful to my oncologist to get her bone marrow biopsy results to share with him?

One case of multiple myeloma (especially if it occurred age 50+) would not be overly worrisome to your health. You can definitely inform your primary care doctor to keep in your family history records but it otherwise wouldn't change your care right now.

Do you have a good recipe for low platelets? Currently 77 and feeling fine! No symptoms, but diagnosed with MDS in February 2017!

With a platelet count of 77 and no bleeding, we would not usually intervene just for the platelet count number. Your MDS doctor can advise you if other intervention is needed for your current MDS based on your whole health and MDS picture.

When it comes to testing your children to see if they have the BRCA1 OR BRCA2 gene, I have been told that if the father is tested and his test comes back negative, his children would not have these bad genes. Is this true?

That is true if the mother's side is not concerning and/or the mother has also had testing. If there is a BRCA1 or BRCA2 mutation in relatives on the dad's side and dad is negative, he cannot pass it on to his kids (that only leaves their mother's side as a potential source and if that side has no cancer, we wouldn't test unless from an ethnic background with higher chance of carrying like Ashkenazi Jewish).

My dad was diagnosed with MDS at 85 and mom with multiple myeloma at 82. What specialist should I seek for genetic testing?

If your mother and father happen to be genetically/blood related OR there were others on either side with blood cancers, then it could be something to look into. Otherwise, both of your parents developed these blood disorders at the typical age and one case alone on each side of the family would not warrant additional major concern or need for evaluation.

If you get MDS at 79 - is there a genetic affect?

Most MDS cases have acquired genetic changes; few diagnosed at age 79 will have inherited genetic changes (if there is a family history of others with MDS or blood count issues, could consider work-up).

How or should I (son) get genetic testing? There is family history of MDS, Prostate Cancer (father), thyroid cancer (sister), breast cancer (maternal grandmother). If genetic trait is found what are the next steps?

The first step is getting to see a cancer genetic counselor or cancer risk program. They will evaluate the details of your family history (exact cancer, age that it occurred, etc.) and help you understand if genetic testing is recommended and what type of testing. The genetic results and your family history and personal risk factors are then used to come up with the best plan for screening and prevention for you.

If a parent had Multiple Myeloma could it be a connection to their 58 yr old daughter being diagnosed with MDS?

Possibly. There are genes such as DDX41 that can be passed in a family and cause both of these cancers.

I have CMML, my dad died of small cell lung cancer and there are other solid cancers in his family, could this be related to my MDS. I am the person with gata 2 p161a.
Small cell lung cancer is very strongly associated with smoking (so far it is not known that that cancer is familial and is more often attributed to smoking). The details of the family history are key to advising regarding any connection to CMML. I don't think the GATA2 P161A explains this full family history.