

Dr. Colleen Jackson-Cook is the leading researcher in mosaic Down syndrome and conducts her work at Virginia Commonwealth University in Richmond, Virginia, USA. On April 1, 2006 we had the honor of speaking with Dr. Jackson-Cook and learning from her expertise on mosaic Down syndrome.

Q. Why is there not a lot of info on MTDS? IMDSA is the only place it is mentioned.

A. ColleenJCook: Largely because it is a fairly uncommon condition. Only about 1-2% of people who have Down syndrome has mosaicism. Given that rarity, there have been limited opportunities to study a large number of people.

Q. Is there any known difference between MDS and MTDS? My husband and I tested negative as carriers.

A. ColleenJCook: Mosaic Translocation Down Syndrome for the most part there has not been any clear predictive difference between trisomy and translocation (mosaic or non-mosaic). However, there are some studies (including ours) that are looking at this.

Q. We recently had a question on our board from a pregnant mom who had an amnio the doctors found Down syndrome in 3 places. What exactly does that mean?

A. ColleenJCook: From that description I can not be certain. I can only guess that they may have found 3 independent clones that had trisomy 21 of the 15 required for study (minimum).

Q. So, is that a percentage estimate then?

A. ColleenJCook: When a prenatal test is done a minimum of 15 independent clones (cells that grew from a single cell) or 20 total cells are scored. I really cannot say if this is a percentage or not without more information they may have the number of 3 places from looking at both amniotic fluid, fetal blood, and possibly placenta. I just cannot know without more info. Sorry

Q. My daughter takes Concerta to help her concentrate to get her homework done, is it real common for MDS kids to have ADD

A. ColleenJCook: I really am not an expert on behavioral issue in Down syndrome, but I have heard several cases of ADD and know it is not an uncommon complication.

Q. My daughter was born with lots of small defects that prompted the NICU doctor to do genetic testing. Most of these were not explained by the MDS diagnosis. Is this common?

A. ColleenJCook: In general, Down syndrome is a condition that has been associated with over 80 clinical findings. Not all findings are present in everyone, and in MDS there is even greater variation. I would need to be able to review the specific situation to answer more thoroughly. If you like you could email me the particulars.

Q. Have you seen brachial clefts with MDS?

A. ColleenJCook: Yes, clefts have been observed in people having Down syndrome.

Q. What are brachial clefts?

A. ColleenJCook: Clefts are areas (usually in roof of the mouth) that do not completely close during development. They sometimes need surgery to close them and can lead to feeding problems, etc.

Q. My daughter's clefts are in her neck and she has severe feeding issues

A. ColleenJCook: I am sorry to hear of her problems. Do you feel that she is getting good resolution of her health concerns?

Q. Yes I do but she has so many unknowns about her. The NICU called her a mystery.

A.ColleenJCook: Has she been seen by a geneticist?

Q. Yes at Children's Hospital Boston.

Q. Based on the percentage a child has, can we make any predictions about the child's level of functioning?

A.ColleenJCook: Great question! We are looking at that as one of our main goals. In general there is a trend toward more severe health problems with higher % of cells. Interestingly, we are also seeing some predictive trends starting to emerge when we use the blood cell % versus the cheek-rubbing %. As soon as this becomes clearer we will let you know, but in general the blood seems to be a better predictor of heart problems; cheek others

Q. Is it too late for me to turn in my research data and photos for my daughter?

A. ColleenJCook: We would love to have them. As we send out result letters we will be requesting additional information that we have not yet obtained or that we have since identified as being helpful for the study.

Q. I am very excited to hear what you have found new

A. ColleenJCook: In general, we are continuing the study and would love to gain any information from people (as well as enroll new ones).

A.ColleenJCook: As an update, we are hoping to analyze data from our study and prepare a paper for publication late in the summer or early fall. Once that has been accepted for publication I will forward to this group.

Q. That will be very helpful Dr. Jackson-Cook. Thank you!

Q. Have you found any correlation age of parents when they had their child with MDS?

A.ColleenJCook: Yes. Using our DNA marker studies we are seeing that in families where the chromosome sorting problem starts in the egg/sperm and is followed by a second sorting problem in the embryo, the parents have an older age compared to cases that have a single chromosome sorting problem in the embryo.

Q. Thanks, I find that interesting, as I was relatively young when my son was born! 24 years old.

A. ColleenJCook: Yes. It is also helpful to remember that while Down syndrome is associated with advanced age, there are many young parents who have a child with DS or MDS since younger people have more children.

Q. My son's initial test of his chromosomes showed some markers. Should my husband and I be tested to see if we carry the gene

A.ColleenJCook: I would need to review the specific chromosome information that was seen in your son to be able to answer this. Could you give me contact information (do we have the original report)?

Q. Yes I do have the original report. I can scan it and send it to you.

A. ColleenJCook: That would be great, or you could FAX it to me. For everyone, if you need to reach me my email is jacksonc@mail2.vcu.edu and FAX number is 804-828-4242.

Q. When I first found out about my son I saw a lady at Children's Hospital in Austin and the lady told me he would never go past the age of 10 mentally, never drive, never live on his own and then talked to about adoption. Where do they get their information? So many people yesterday had that same experience and all of them went to different doctors

A. ColleenJCook: Yes, it is unfortunate that people often do not have updated expectations. Much of the old data is based on cases that were raised in institutions and did not have educational opportunities, etc that exist today. As for your son's progress, each child is different and we just have to help each one to attain their best (just like any other child).

Q. Are there any known environmental causes of MDS? My husband works with pesticides and I am an oncology nurse. Also my husband just found out that someone he used to work with had a child with MDS 5 years ago.

A. ColleenJCook: There have been many studies to try to identify environmental causes of DS and MDS. To date, none have been identified in humans. Part of the problem is that the environmental exposures are so varied and the frequency of chromosome problems makes a clear association difficult to identify. Even the atomic bomb and the Chernobyl accident could not be clearly associated with any increase in frequency. I want to make sure it is clear that the pesticide exposures, etc are NOT thought to be a cause of chromosome sorting problems.

Q. With XYY it's hard for us to know what is affecting Jason. He seems to have few MDS issues & I've been told that kids can "out grow" issues related to XYY. Is that true?

A. ColleenJCook: I really do not know that people "outgrow" their issues with XYY. As they mature they may be able to better control impulsiveness, but some do perform better with intervention training (and medication in some cases).

Q. I had hepatitis A and miscarried when I was 2 months pregnant. Then a month later I got pregnant with my daughter who has MDS. Is there any correlation?

A. ColleenJCook: I am not aware of any correlation between hepatitis A and MDS. Many viruses have been studied as potentially causes for chromosome problems and no clear association has been identified.

Q. Which sample, blood or cheek is the best predictor of % in brain cells?

A. ColleenJCook: We still do not know the answer to that question. To try to better assess this, we will be asking for developmental information of people who participated in the study (if they are willing to share) to see if we can gain insight.

Thank you Dr. Jackson-Cook for taking the time to answer our questions. I think that we have all learned a lot tonight and each of us has a better understanding of mosaic Down syndrome.

If you would like to participate in the mosaic Down syndrome research conducted at Virginia Commonwealth University, you can contact Dr. Colleen Jackson-Cook at:

Colleen Jackson-Cook, PhD
Department of Human Genetics
Virginia Commonwealth University
P.O. Box 980033
Richmond, VA 23298-0033
Phone: (804) 828-9632 ext. 118
Fax: (804) 828-3760
E-mail address: JACKSONC@HSC.VCU.EDU

You can also read her research study at our web site at: <http://www.imdsa.com/VCU.htm>