



## FA Connect: Question & Answer Transcript

**Session Title:** In vitro fertilization (IVF) with preimplantation genetic testing (PGT) for Fanconi anemia and human leukocyte antigen (HLA) matching

**Invited Speaker:** Dr. Ilan Tur-Kaspa, Founder & Medical Director of the Institute for Human Reproduction (IHR)

**Session Air Date:** January 12, 2021

Access a recording of Dr. Tur-Kaspa's presentation by visiting [www.fanconi.org](http://www.fanconi.org). The recording has been uploaded with Spanish subtitles.

Have additional questions for the speaker? Email Dr. Tur-Kaspa directly at [drTK@infertilityIHR.com](mailto:drTK@infertilityIHR.com).

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**Q:** I'm a patient with Fanconi anemia (FA) but I'm in early menopause. We want my sister to be the egg donor, but we don't really know if she's a carrier for FA. What should the test look like to make sure she's not a carrier and/or has any specific FA genes?

**A:** This would be done through genetic testing, same as how you, an individual with FA, were tested. Your sister should go in for genetic testing, and so should your husband. If he is not the carrier and she is not a carrier, then the child will be born without FA.

**Q:** What is the approximate cost to families when they go through this process?

**A:** It depends. Some will have insurance that may cover the process. The IVF as well as the PGT could be covered if you do it for PGT-m. They may not necessarily cover the PGT-a that we add to the mixture or the HLA, but usually if the insurance covers PGT-m, I think that they will also cover the PGT-HLA. However, they will not cover the PGT aneuploidy; this is an additional cost of \$2-3,000. **Altogether, IVF cycle in the US may cost around \$12-15,000.**

We usually provide some discount for families that come to us because we understand that unfortunately, some of them may need to have another 1,2,3 or 4 cycles in order to get an HLA-match embryo. In our labs we perform the embryo biopsy and embryo cryopreservation. As for the PGT lab, now we're looking at cost for the PGT-m, PGT-HLA and PGT aneuploidy. Altogether, that can be \$5-10,000 per cycle. The assays may also have a cost which can vary depending on the age of the woman and how she responds to the medications. Altogether, it can vary between about \$3-6,000 just for medication.

There may also be some travel costs involved. So, yes, it may be costly each cycle. Before we begin the process, we will explain exactly how much the process will cost, and they will get these figures in writing. I wish we could do everything for free, but unfortunately, this is not the situation. We try to help as much as we can.

**Q:** I'm wondering what the upper age limit is for a couple who would like to go through this process?

**A:** Sooner than later. In human reproduction, statistically, when someone is less than 35 years old, the chance to conceive with IVF per cycle is 22%. It's not 80% or 90% or 70%; it's 22% each time we try. And yes, most couples who don't suffer from infertility will conceive within a year. They will have around an 85% chance to conceive within that year. Again, this is for those less than 35 years of age. Once you are over 35, the chance to conceive starts to go down. By 38, it's at 15%, by 40 maybe around 10%, over 42 is less than 5%, and less than 1-2% at 43 or 44-year-olds. In addition, the chance to have a miscarriage, significantly increases with age. If you're less than 35 and you are pregnant, the chance for a miscarriage is around 15-20%. The chance for a miscarriage if you're 42-43 goes to 50%. So now, the chance to conceive at that age may only be up to 5%, and the chance for miscarriage is around 50%.

We have some women who are 26 years old and have diminished ovarian reserve. While they may look younger than their age, their ovaries may respond like they're 43-years-old already. We also have women who are 42 or 43 that look younger than their age, but their ovaries will respond like they are 38. Altogether, it is so important to remember that the chance to conceive decreases significantly with age.

**When we talk about HLA matching embryos, it's even more important because only one in four will be an HLA match. If you're doing this for Fanconi anemia, it's three out of four. So now we're talking about three out of 16 embryos will potentially be an HLA-match. If you also take the aneuploidy rate (being about 50% +/- depending on age) out of those embryos, maybe one or two out of 16 will be normal for chromosomes, unaffected by Fanconi anemia and an HLA-match.**

So altogether, if you are 41-42, I can tell you that if you have 1-2 blastocysts that we can do biopsies and tests, that's fantastic. Out of those, I have 1 or 2 out of 16 that will be HLA-matched and without FA. Even if you don't want to conceive now, get the embryos on ice. Bank your embryos, because if you come to me years later when you are 44 or 45 without doing so, I'll have to tell you that the chance to conceive from your own eggs is extremely low; less than 1-2%, or an almost zero chance of having a natural egg match embryo. If you have frozen embryos, even at 45 or even 55yo, you'll have a much better chance of becoming pregnant.

**Q:** There is some travel involved with this process. I'm wondering if some of this process can be done locally and/or how often do families have to go to Chicago for your services?

We have established excellent working relationships with centers all over the world. People from the Netherlands or Australia, for instance, can collaborate with IHR through a local center and I will do the monitoring, the medication, IVF, can be done there at home. Once we receive the results and review them, we contact the family's local center and provide the orders to continue.

We ask families to travel to Chicago and arrive two to four days before the planned egg retrieval. Final monitoring is done in Chicago to evaluate the ovaries, provide the final trigger shot with egg retrieval at that time. The family can fly home the next day, while we await the genetic testing results. Once we have an embryo that is good for transfer, we plan for the next step. In addition to frozen embryo transfer, we may do a "mock cycle" for the endometrial receptivity assessment (ERA) test, in order to also concentrate on the uterine cavity and to improve the receptivity as best as possible. Multi-center perspective studies show that the ERA test may improve the chance to conceive with frozen embryo transfer by 15%. The embryos that we have are very precious. We want to optimize the process from day one and try to get as many good eggs (embryos) as we can and want to ensure the uterine cavity is doing well, so as to provide synchronicity between the two of them.

Next is the embryo transfer. One embryo is transferred at a time. The reason being, is if we have an embryo with chromosomes good for Fanconi anemia HLA-match, the chances to conceive per transfer with one embryo may be between 50-70% success rate, depending on the woman's age. Compared to the numbers that I provided earlier, now the success percentage may be very high.

Lastly, when you travel to the US for medical treatment, you do not need to quarantine. You will need to be tested for COVID-19 before you arrive and after you leave, but you will not need to quarantine.

**Q:** I'm calling from Europe. I wondered on average, how long a typical process would take? I was told on average it'll take about one year for the test probe—even before you start the IVF. Is that correct?

**A:** In Chicago, the probe will be done within 3-6 weeks, and you can start the treatment in the following 4-8 weeks (depending on your menstrual cycle). We are very fast, but my philosophy has always been that if asked when you'd like to start, you'll typically respond "yesterday". Therefore, it's our job to make sure that it happens quickly.

Unfortunately, in Europe, there are still many centers that take a long time to prepare the probes, etc. From a genetic point of view, if someone starts working on it tomorrow, it will take a few weeks. This is just a matter of priority and resources, whether it's private or social medicine.

I cannot apologize for everyone, but if you contact us (IHR), once we have the genetic materials for testing and setup for the whole family, the process can be completed in 3-6 weeks and we can then start IVF. If you come to me, I can tell you that within a year, you could deliver—not just start.

There are still some countries that have some ethical restrictions and do not allow this process at all. From our point of view this process is ethical.

**Q:** What if you have small children and it is difficult to travel to the US for this treatment?

**A:** We can have the sperm frozen and shipped to the US, so if you need to come alone for treatment, your partner can stay with the kids at home. You could also come out for a week or two, but your husband could just come for 3-4 days to do two things: first, to provide sperm on the day of the egg retrieval, and second, to take care of you after the retrieval.