

Interview

Acute myeloid leukemia patient genome interview



The anonymous patient (AML2) who took part in this interview is one of approximately 180 patients that Dr Timothy J Ley from the Washington University School of Medicine (St. Louis, MO, USA) and his leukemia colleagues accrued for their study investigating the genetics of acute myeloid leukemia (AML) from the beginning of the year 2000. His team of researchers used a fledgling PCR-based re-sequencing pipeline to target and sequence various suspect/candidate genes in AML. However, this search for new genes that are frequently mutated in AML proved to be largely unsuccessful and led to the notion of whole-genome re-sequencing (e.g., that of an unbiased look at all genes and structural changes). Therefore, the sequencing of this interviewee's genome followed the researchers' first AML genome case, published in *Nature* on 6 November 2008 [1], and since the first patient that they sequenced was a female (now deceased of her disease), they decided to choose a male patient in remission from the disease but with the same AML subtype (M1). Hence, AML2 was sequenced and analyzed – they identified recurring mutations that may be relevant for the pathogenesis of the disease. They subsequently published their results in the *New England Journal of Medicine* on 10 September 2009 [2]. The ultimate goal of their research is to change the 'standard of care' for AML patients, since for the last 25 years, doctors have been treating every AML patient in basically the same way, never knowing who will respond and go into remission, or who will not respond and will need a bone marrow transplant.



■ **What made you decide to participate in the research that sequenced your entire genome?**

The doctor had spoken with me about it from the beginning and had basically told me that it was just a study that might help future patients and that it wouldn't affect me one way or another as far as my treatment or anything of that nature was concerned. So I thought if it could help somebody else, I would be all for it and would not have a problem with it.

■ **Could you briefly summarize your experiences of having your genome sequenced?**

Well, I really didn't think about it very much until I found out the results just recently. I knew that the study was going on but it really didn't affect me in any way as I didn't think about it very much. It didn't really come up in discussion too often either so it didn't make a difference to me one way or another.

■ **How did your family feel about you participating in this study?**

My wife was ok with it and I think I had spoken to my mom about it too – it really

wasn't discussed a whole lot – it was just a procedure that I told them I didn't need to be involved in but that I had volunteered to do it. I had no idea that the results would lead to where it's at now. They seemed ok with it.

■ **Could you briefly describe how they went about taking a sample of your DNA?**

From what I remember, it happened while they were taking a biopsy from my bone marrow. They first took the [biopsy] sample and after that procedure was finished, they explained that they were then going to take the sample for the study. I am not sure what instrument they used though – the area was already numb and I couldn't really feel a whole lot at the time. It didn't last very long and it wasn't that much of a big issue to me.

■ **Was taking a sample of your DNA painful?**

No, not at all. It was just something that was part of the normal procedures that I was undergoing at the time with my treatment.



■ **How did you feel about having a sample taken?**

Well, at the time I felt good about it, knowing that I would hopefully be able to help somebody else out and [I] hoped that anything they discovered from the study would help [patients] in the future. I was certainly surprised when all the results were released recently – it was a good feeling!

■ **What were the advantages/disadvantages of taking part in the study?**

I can't think of any disadvantages.

The advantage (I felt at the time when the study first started) was that if I was able to help somebody else, I was more than willing to participate. Then, after the results had come back (after they completed the testing and compared my results with the other samples in the study), the other advantage would be that I was glad they were able to do it [the study] and it is just a matter of another step towards helping people out in the future. So it just made me feel good to be able to help them out.

■ **How did getting the results back affect you?**

I was in shock at first, and just to think (from what I was told) that I am only the second person in the history of mankind

that they were able to do this with – I felt blessed.

■ **What is your opinion about personal genomics since you have actually had your entire genome sequenced?**

Oh, I feel that it's a good thing! It is just amazing what they are able to do now, I had no idea that it [the research] would be able to lead to this. From what I understand from reading about it now, that is, what had been performed and how the results come out, it sounds like it's going to be getting bigger and better and more people are going to be able to benefit from this [technology].

■ **Do you think this type of research will help improve the 'standard of care' that is currently given to acute myeloid leukemia patients?**

Yes, I think so. I think we will have a better understanding and hopefully be able to develop better treatments. From what I have been told since I was first hospitalized, treatments have improved drastically from when patients had the disease 10–15 years ago compared with when I was first diagnosed. It's amazing to think that in the last few years they have gotten this far! It is just getting better each day and I think it will certainly help in the future and I will support it 100%.

References

- 1 Ley TJ, Mardis ER, Ding L *et al.*: DNA sequencing of a cytogenetically normal acute myeloid leukemia genome. *Nature* 456(7218), 66–72 (2008).
- 2 Mardis ER, Ding L, Dooling DJ *et al.*: Recurring mutations found by sequencing an acute myeloid leukemia genome. *N. Engl. J. Med.* 361(11), 1058–1066 (2009).