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Dear RUNX1 Natural History Study Participant,

Time is flying by these days! Even with a brief slowdown in patient visits to the NIH due to the Omicron variant of COVID-19, our team has been staying busy. When we're not taking care of our research participants in-person or virtually, we've been hard at work analyzing all of the data we collect with the help of research participants like you.

We are almost ready to submit two manuscripts outlining our findings for publication so that other providers and scientists can learn from you. The first will outline all of the clinical information you've taught us — like new symptoms of RUNX1 beyond bleeding, bruising and risk of blood cancers — and will provide better statistics on how likely patients are to develop cancers.

Our second paper will dive into our genetic and genomic studies. This paper details all of the different ways we've seen the RUNX1 gene be "misspelled" and will also outline some of the secondary somatic variants (non-inherited genetic changes that affect only a few cells) that have been detected in our cohort. Understanding these somatic variants will be important for us to understand which people with RUNX1 are at a higher or lower risk of developing cancer.

We hope these two papers are the first of many to come out of this study. Getting them out in the world will be essential to understanding this disease and working to find better treatments for the RUNX1 community.

Study Statistics So Far

- 171 individuals with RUNX1 from 71 families
- 69 participants evaluated at the clinical center
- 98 unaffected family members
- Participants from all different ages (0-75 years old)
- More than 20,000 pages of medical records obtained
- 50 unique variants in the *RUNX1* gene
- Exome data on bone marrow from 128 patients
- Genomes data (even bigger than an exome!) on 7 patients

Changes to Our Team

As always our team is changing.

We recently welcomed Shawn Chong, PA-C, to the team. He is the primary patient care provider, including performing bone marrow and skin biopsies. We are especially excited about this because it will give us additional scheduling flexibility by not having to rely on other NIH providers to do procedures.

We are also saying goodbye (but only partially) to Dr. Lea Cunningham. Dr. Cunningham will focus her time on her role with the NIH Bone Marrow Transplant Program and will be working to develop treatment trials for individuals with RUNX1. We are thankful for the time that Dr. Cunningham has spent with the team so far, and we look forward to having her as an important collaborator moving forward.

Dr. David Young, a board-certified pediatric hematologist from NHLBI, has joined the team as the medical advisory investigator to support our patients from a hematologic perspective.

We are also ecstatic to welcome our research nurse Katie Craft, BSN, RN, back to the team. Katie is a familiar face to many of our patients and has been very much missed!

COVID-19 and the Natural History Study

Things are starting to get back to almost normal at the NIH Clinical Center, and we are now able to see most of our U.S.-based patients just as we would have before the pandemic. If you would like to come into the NIH and are not already in the process of being scheduled, please reach out to Shawn Chong (shawn.chong@nih.gov) or runx1@nih.gov.

Unfortunately, we are still unable to bring in international participants, but we hope this will change soon. While we are not able to bring international patients to the NIH, we do have options for remote enrollment available.

In the meantime, the RUNX1 team is always a click away, and we are always available to answer any of your questions or concerns virtually.

Options for Participating in the RUNX1 Natural History Study

Understanding RUNX1 will take enrolling as many participants as possible; however, we also understand that everyone has busy lives outside of having RUNX1, so we want to offer as many options as possible for participating in our study. If you or other members of your family are interested in joining the study but are unsure if you have the time to commit to the full work, some of these options may be helpful for you.

1. **Annual NIH Clinical Center visit for 1.5 - 4 days.** The shortest option would be available to patients who are

able to tolerate procedures done with local anesthesia. The longer option allows patients to have additional subspecialty consultations to provide more detailed and consistent data for the study and provide them with more tailored results and care.

2. **Remote enrollment with a telehealth visits with our team.** We would ask for patients to work with their home care provider to get certain clinical labs drawn and to share data from any procedures they may receive at home. We would also ask that patients send a few research samples to our team for analysis in our lab. We would then be able to meet via telehealth and go over the patient's clinical history and any recommendations, just as we would if the patient came to the NIH. In some cases, we may also ask for the patient to meet with our consulting teams such as allergy and immunology over telehealth.
3. **Remote enrollment.** This is the most hands-off approach. The patient would be able to share you're their medical records with us through a secure link, and we will arrange for a remote blood draw for our research level tests approximately once a year. The consent can be done electronically.

We often say that this is a "choose your own adventure" clinic. We are happy to work with you to come up with the best plan for your participation in the study using any combination of the options above. It's whatever fits the best for your family.

NIH Patient Meeting Update

While we had hoped to have a patient meeting at the NIH in 2022, we have decided to postpone it indefinitely due to COVID and logistics at the Clinical Center. Our team will attend the RUNX1 Research Program's scientific and patient meetings, so we will hope to see many of you virtually or in Princeton, New Jersey, in the fall!

Rare Disease Day

On February 28 our team celebrated Rare Disease Day by donning our best zebra gear and tuning into some amazing talks from NIH researchers. The NIH Rare Disease Day virtual conference was full of important talks and stories from researchers and patients with rare diseases. We hope that

many of you all were also able to tune in, but if you did not recordings are available at <https://ncats.nih.gov/news/events/rdd>. More information on global Rare Disease Day events can be found at <https://www.rarediseaseday.org/>.

If you didn't already know, Rare Disease Day is technically the 29 of February as it's the rarest day of the year; however, because there was no February 29 this year, we celebrated on February 28. And if you're wondering, "What's with the zebras?" We love zebra's in the rare disease community. It's a nod to the medical adage, "When you hear hoof beats, think horses not zebras." However, we know that our patients with rare diseases are the zebras that are so often missed by the medical community.

Accessing Your NIH Medical Record

As a reminder, you can access clinical results from our study through the NIH FollowMyHealth Patient Portal.

Information about setting up your patient portal can be found here: <https://clinicalcenter.nih.gov/followmyhealth/index.html>

Thank you for your participation in the NIH RUNX1 Study!
Please contact us with any questions!

Sincerely,
The RUNX1 Study Team