

TRACK-FA NEWSLETTER

ISSUE 5 – AUGUST

2024



TRACK-FA is a longitudinal natural history study that tracks brain and spinal cord changes in individuals with Friedrich's ataxia (FA). We have an outstanding team of researchers from Australia, USA, Germany, Brazil and Canada and we collaborate with global industry partners. Our goal is to investigate the sensitivity of neuroimaging biomarkers to provide a basis to include them in future clinical trials.

AT A GLANCE

As of August 2024, we are rapidly approaching the end of Study Visit 2. Our current recruitment numbers are presented in Figure 1.

- 229 participants have completed Study Visit 2.
- 107 participants have completed Study Visit 3.

For each visit, we collect:

- MRI scans
- clinical assessments
- speech assessments
- cognitive and mood assessments
- biospecimens

We are on track to complete:

- Study Visit 2 assessments by October 2024
- Study Visit 3 assessments by October 2025

We thank everyone who continues to participate in TRACK-FA and contributes to this important dataset!

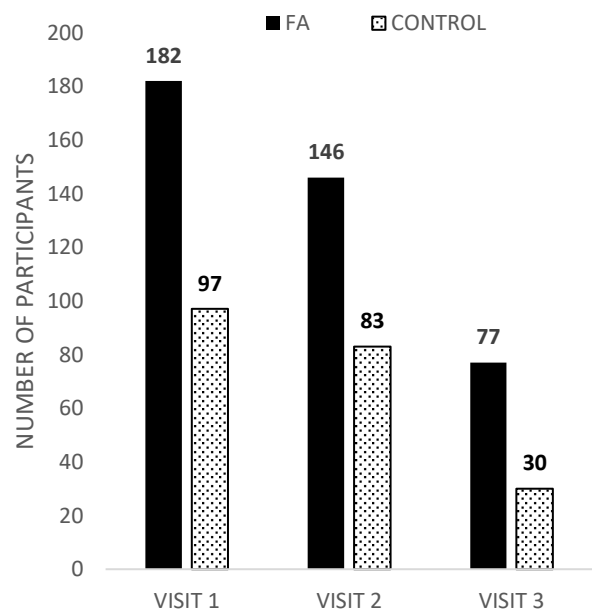


FIGURE 1. NUMBER OF TRACK-FA PARTICIPANTS WHO HAVE COMPLETED STUDY VISITS 1, 2 AND 3 AS OF AUGUST 2024.

RESULTS UPDATE – WHAT HAVE WE FOUND SO FAR?

We are preparing a publication to present our findings from the baseline analyses (Study Visit 1) of the TRACK-FA cohort. Below is a preview of some of our preliminary results.

While many of our measurements show similarities between individuals with FA and individuals without FA (the control group), we focus on the regions that show *differences* between groups as these may be the most useful biomarkers to track disease progression. Figure 2 shows some of the main differences between individuals with FA compared to individuals without FA.

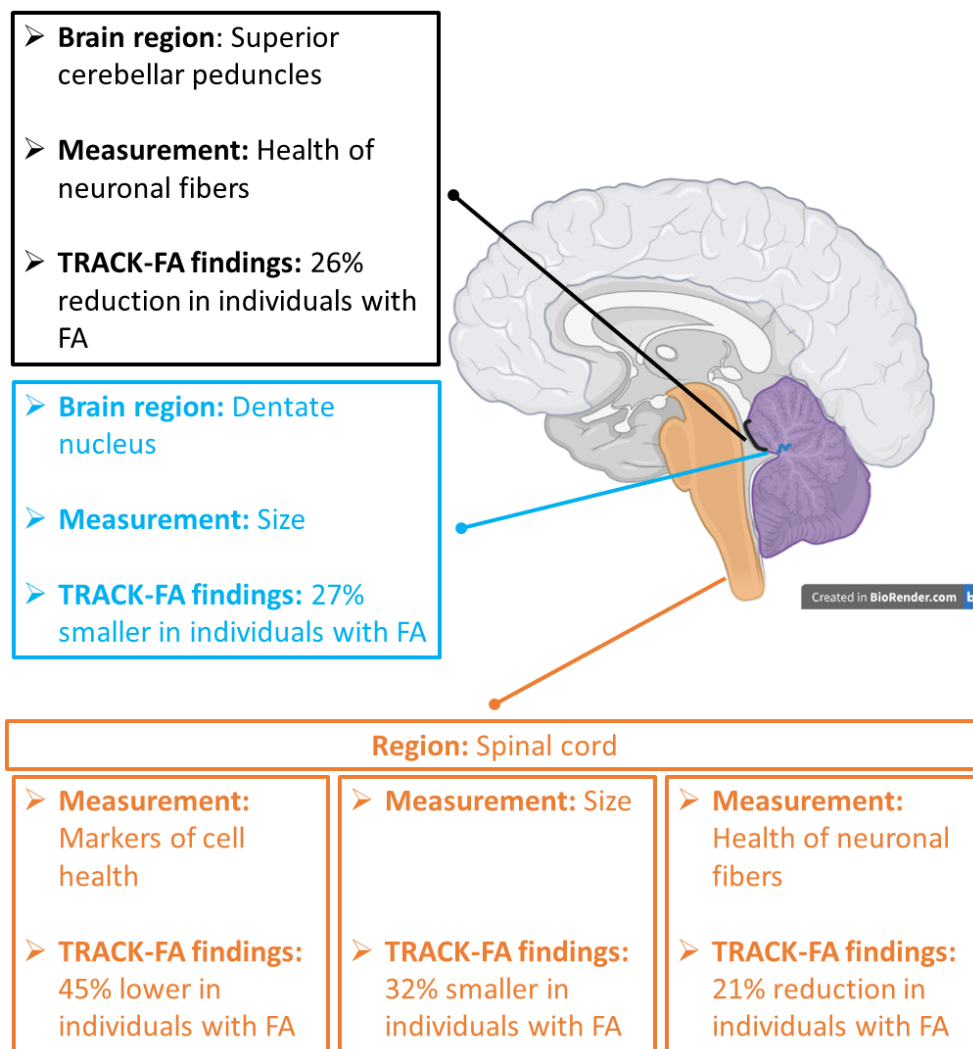


FIGURE 2. BRAIN AND SPINAL CORD DIFFERENCES IN INDIVIDUALS WITH FA COMPARED TO INDIVIDUALS WITHOUT FA.

Why are these finding important?

We know these areas play a significant role in movement, sensation and balance.

Understanding how FA affects these areas will help researchers to identify if current or future therapies will assist in improving connections between the brain and spinal cord, or if they help increase the size of some parts of the brain and spinal cord.

Which TRACK-FA measurements show differences between individuals with and without FA even from childhood?

TRACK-FA is the first multi-modal neuroimaging study to encompass both children and adults, spanning 5 to over 40 years of age. The TRACK-FA dataset includes very young participants with FA, with approximately 10% of participants aged 10 years or younger. This allows us to identify which differences are present even from very early in the disease.

By looking at the TRACK-FA data from Visit 1 and comparing individuals with FA to individuals without FA, early results show:

- For some measurements – such as the size of the spinal cord – **differences can be detected even in very young children**, and become more different as age increases, as shown in Figure 3.
- For other measurements in the brain, **differences can be detected later, in older children or adults.**

Why are these findings important?

Examining how these differences change over time in subsequent visits will be very important. This will help determine if these differences can serve as effective outcome measures for clinical trials involving children.

Spinal cord size as age increases for individuals with FA compared to individuals without FA

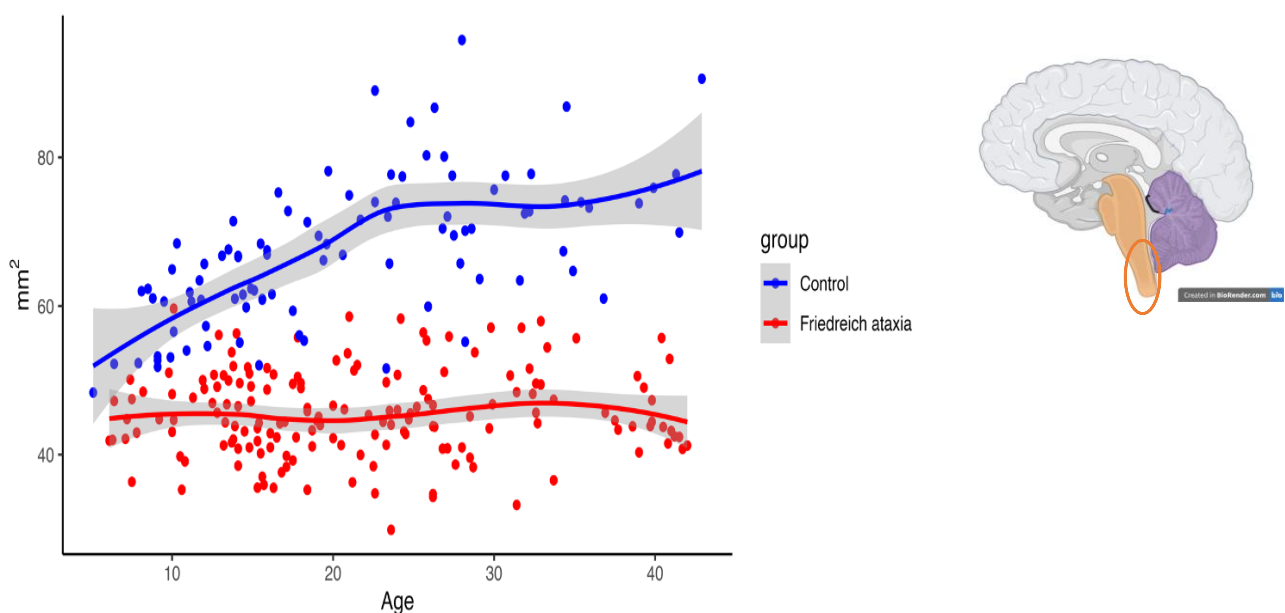


FIGURE 3. SIZE (CROSS-SECTIONAL AREA) OF THE CERVICAL SPINAL CORD IN INDIVIDUALS WITH AND WITHOUT FA DEPENDING ON AGE. Results show the size of the spinal cord increases steadily during childhood in individuals without FA, but does not increase with age in individuals with FA.

MEET OUR TRACK-FA TEAM MEMBERS



Name: Professor Pierre-Gilles Henry

TRACK-FA Site: University of Minnesota (Minneapolis, Minnesota, USA)

Role in TRACK-FA:

I am the TRACK-FA coordinating Principal Investigator (PI) for neuroimaging, and, together with Christophe Lenglet, PI for the University of Minnesota site. My main role is to coordinate all imaging activities across all seven sites to obtain the best imaging data possible. TRACK-FA is a large team effort!

Highlights of TRACK-FA so far:

What's special with TRACK-FA is that, for the first time, we are gathering large amounts of data that cover the entire age range from young children to teenagers to adults: the age of TRACK-FA participants ranges for 5 to 55 years old. Neuroimaging data from MRI show how development of the nervous system in FA differs from normal development. For example, we see that, in FA, most of the brain develops normally, but the spinal cord and brain stem (which connects the spinal cord to the brain) do not seem to fully develop. This is crucial information for pharmaceutical companies developing potential treatments.

What have you learned from being part of TRACK-FA?

I have learned what is possible when many talented people across four continents join forces towards a shared goal. From the beginning, there was a strong desire from researchers to collaborate and combine our expertise to achieve something big and make a difference in FA. I also learned how important it is to have an organization like FARA that brings the community together and catalyzes research efforts and fundraising. TRACK-FA would not have happened without FARA!

What have you learned from TRACK-FA participants?

I started working on FA in 2013 and was immediately struck by how special the FA community is. I really enjoy meeting study participants and their families. The FA community is very active in participating in research.

Tell us about yourself.

I enjoy skiing, swimming, running, and generally being in nature. I like traveling in different countries, and getting to know other cultures and languages. Over time I have learned several languages including German, Japanese, and American Sign Language, and I am fluent in two: French (my native language) and English (with a French accent!). I also greatly enjoy reading: two of my favorite novels are 1Q84 by Haruki Murakami, and The Grapes of Wrath by John Steinbeck.



Name: Dr Sandro Romanzetti

TRACK-FA Site: University
Clinic RWTH Aachen, Germany

Role in TRACK-FA:

MR physicist

Highlights of TRACK-FA so far:

Every time a patient steps into our Lab, it brings a smile to my face. Their presence brightens my day, and I'm grateful for the chance to assist them. Equally fulfilling is the unique opportunity we have to delve into the complexities of this challenging disease. By delving deeper into our research and making progress in our understanding, we are taking important steps toward improving the lives of many people. Our goal is to make their lives better and provide them with the hope for a brighter future.

What have you learned from being part of TRACK-FA?

Working alongside numerous talented scientists worldwide not only enhances the quality of our work but also affords us the invaluable opportunity to learn from their diverse expertise. The collective wealth of knowledge within our global network of scientists is truly astonishing, enriching our endeavors and fostering continuous growth.

What have you learned from TRACK-FA participants?

Remarkably, every patient enthusiastically embraces their participation, unwavering in their dedication, even during lengthy visits. Their commitment to advancing our understanding of FA is unwavering, underlining their invaluable contributions to the scientific community. It's worth noting that these patients are never alone; their families, equally wonderful and friendly, consistently go above and beyond to play an integral role in this study.

How did you get involved in FA research?

In my role as an MR-Physicist, I've been actively engaged in various clinical studies encompassing a range of neurodegenerative conditions. Yet, my deep interest was triggered by the distinct patterns of degeneration specific to FA. The spinal cord and cerebellum, being pivotal areas, have captured our attention, motivating us to employ our most advanced MRI techniques in the quest to extract comprehensive insights from these regions.

Tell us about yourself.

In my free time, I thoroughly enjoy spending quality time with my son, Giorgio, who has an immense passion for fast trains. Whenever we get the chance, we embark on day trips to explore nearby cities together. Additionally, my heart lies in the world of tennis, where I find joy both in playing and sharing my expertise by coaching at my local club.

CONTACT YOUR LOCAL TRACK-FA SITE

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<p>Philadelphia, Pennsylvania, USA Children's Hospital of Philadelphia, Philadelphia, USA Study contact: Willa Mazullo E: mazullo@chop.edu T: 267-425-4150</p> 	<p>For more information about the TRACK-FA Study, see our website</p>  <p>https://www.monash.edu/medicine/trackfa</p>

Further information

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