

Natalia Acosta-Baena: The genetic gap between neurodevelopment and neurodegeneration

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Dr. Natalia Acosta-Baena embodies the rare confluence of clinical and basic science expertise that modern neuroscience demands. As a physician-scientist at the University of Antioquia's Neurosciences Group, she combines her medical training with a master's in clinical epidemiology and doctoral studies in basic biomedical sciences, specializing in Genetics. Her groundbreaking work began with contributing to characterize the world's largest population affected by autosomal dominant genetic Alzheimer's disease (mutation E280A in PSEN1), establishing a foundation for numerous studies on biomarkers, clinical trial design, and genetic modifiers in this pivotal cohort. In a landmark discovery, Dr. Acosta-Baena's research revealed a novel syndrome linked to a SPAG9 variant, demonstrating how a single gene involved in neuronal retrograde transport can drive neurodevelopmental problems and neurodegeneration in affected patients. This finding challenges the traditional separation between these processes and suggests shared biological pathways. Through her continued work with families affected by neurodevelopmental disorders, she has uncovered genetic networks that reshape our understanding of rare brain diseases. Her current translational medicine and genetic epidemiology research focuses on further exploring these unexpected connections between neurodevelopment and neurodegeneration. In a Genomic Press Interview, Dr. Acosta-Baena shared her life beyond the laboratory – from her early fascination with the human brain to finding joy in Colombia's mountain sunsets with her husband and son and drawing inspiration from Latin American writers like Cortázar and García Márquez. Her dedication to scientific rigor and human connection is reflected in her philosophy that each failure teaches something essential as she works toward translating genetic discoveries into meaningful healthcare policies and personalized medicine approaches.

Part 1: Natalia Acosta-Baena – Life and Career

Could you give us a glimpse into your personal history, emphasizing the pivotal moments that first kindled your passion for science?

It may sound strange, but ever since I can remember, I have wanted to study medicine and understand our brain. I do not have parents who were doctors or anyone in my family to influence me. It has motivated my life since I was little. I remember that I didn't like going to school, but I understood that it was the only way to get to work in what I was passionate about.

We would like to know more about your career trajectory leading up to your current role. What defining moments channeled you toward this opportunity?

A motivation: neuroscience research. Two opportunities: I started by attending a study group in Neuroanatomy with the Neuroscience Group of



Figure 1. Natalia Acosta-Baena, MD, MsC, PhD(c), Universidad de Antioquia, Colombia.

the University of Antioquia, and I accepted the opportunity to do a master's degree in epidemiology with the same research group with Professor Francisco Lopera and then complete a doctorate in genetics.

Please share with us what initially piqued your interest in your favorite research or professional focus area.

The complexity of human thought and action and the search for the evolutionary question of what the brain of homo sapiens had genetically and physiologically made us the species that managed to survive above the other hominids.

What impact do you hope to achieve in your field by focusing on specific research topics?

The greatest impact I hope to achieve with my research is to foster a collaborative effort that reaches individuals who are sick and their caregivers. I envision a future where our knowledge empowers people and





influences health policies towards true prevention. Our studies, currently in the form of articles, hold the potential to be applied to the general population, shaping decisions and responses for patients.

Please tell us more about your current scholarly focal points within your chosen field of science?

Neurodevelopmental genetics is a field where we only see the tip of the iceberg. We do not see what we do not understand. When we manage to understand the relationship between genetic networks, we will be able to decipher neurodevelopment and neurodegeneration. My focus is genetics for prevention and personalized and community medicine.

What habits and values did you develop during your academic studies or subsequent postdoctoral experiences that you uphold within your research environment?

Continue with the medical consultation. Each rare clinical presentation of a disease that surprises is what motivates new questions.

At Genomic Press, we prioritize fostering research endeavors based solely on their inherent merit, uninfluenced by geography or the researchers' personal or demographic traits. Are there particular cultural facets within the scientific community that warrant transformative scrutiny, or is there a cause within science that deeply stirs your passions?

I am passionate about new questions and new challenges. I am bored by absolute certainty and those who believe they have it.

What do you most enjoy in your capacity as an academic or research rising star?

That I do not feel like one.

Outside professional confines, how do you prefer to allocate your leisure moments, or conversely, in what manner would you envision spending these moments given a choice?

I always enjoy the view of the sunset or the rain in the mountains of Colombia with my husband and son.

Part 2: Natalia Acosta-Baena – Selected questions from the Proust Questionnaire¹

What is your idea of perfect happiness?

There is not one, both words are a pleonasm.

What is your greatest fear?

"Without music, life would be a mistake" "Ohne Musik wäre das Leben ein Irrtum" (Friedrich Nietzsche, *Die Götzen-Dämmerung – Twilight of the Idols*, section: "Sprüche und Pfeile" (Maxims and Arrows) aphorism #33, 1895).

¹In the late nineteenth century, various questionnaires were a popular diversion designed to discover new things about old friends. What is now known as the 35-question Proust Questionnaire became famous after Marcel Proust's answers to these questions were found and published posthumously. Proust answered the questions twice, at ages 14 and 20. In 2003 Proust's handwritten answers were auctioned off for \$130,000. Multiple other historical and contemporary figures have answered the Proust Questionnaire, including among others Karl Marx, Oscar Wilde, Arthur Conan Doyle, Fernando Pessoa, Stéphane Mallarmé, Paul Cézanne, Vladimir Nabokov, Kazuo Ishiguro, Catherine Deneuve, Sophia Loren, Gina Lollobrigida, Gloria Steinem, Pelé, Valentino, Yoko Ono, Elton John, Martin Scorsese, Pedro Almodóvar, Richard Branson, Jimmy Carter, David Chang, Spike Lee, Hugh Jackman, and Zendaya. The Proust Questionnaire is often used to interview celebrities: the idea is that by answering these questions, an individual will reveal his or her true nature. We have condensed the Proust Questionnaire by reducing the number of questions and slightly rewording some. These curated questions provide insights into the individual's inner world, ranging from notions of happiness and fear to aspirations and inspirations.



Figure 2. Natalia Acosta-Baena and her son embracing the beauty of the Andioquia Mountains during Colombia's COVID-19 lockdown (25 March 2020). Against a backdrop of lush tropical vegetation and bamboo groves characteristic of the region's mountainside, they stand on a cleared hillside slope, arms outstretched in a moment of joy and freedom despite the global pandemic restrictions. The contrast between the cultivated slope in the foreground and the dense forest canopy above captures the typical landscape mosaic of the Andean countryside.

Which living person do you most admire?

To every woman in Latin America who prioritizes her time for the integral education of her children above any other need.

What is your greatest extravagance?

Be happy.

What are you most proud of?

Being a mother.

What is your greatest regret?

I do not regret anything so far.

What is the quality you most admire in people?

Honesty and good humor.

What is the trait you most dislike in people?

Arrogance and prepotency.

What do you consider the most overrated virtue?

None. Each virtue is relevant to humanity, and research on humans and animals is an example.



What is your favorite occupation (or activity)?

Writing – and I can – dancing.

Where would you most like to live?

In Colombia, in my current house in a rural area in the mountains.

What is your most treasured possession?

My memories.

When and where were you happiest? And why were so happy then?

When my son was born, 10 years ago, and all the good times since then.

What is your current state of mind?

I currently feel calm and grateful for everything I have received in life.

What is your most marked characteristic?

Imagination.

Among your talents, which one(s) give(s) you a competitive edge?

I was not born with special talents, but I was born with a great deal of curiosity and a desire to develop new skills every day.

What do you consider your greatest achievement?

Every goal achieved is the biggest at the time. Every article achieved and published is the sum of the efforts of many people, so it is great.

If you could change one thing about yourself, what would it be?

I do not want to change anything. What I was and what I am have left me where I am now, and I feel proud of where I am, how I am, and who I am with.

What do you most value in your friends?

Appreciation and loyalty despite thousands of flaws and mistakes.

Who are your favorite writers?

Julio Cortazar, Horacio Quiroga, Jorge Luis Borges and Gabriel Garcia Marquez.

Who are your heroes of fiction?

None. My favorite is an antihero, the Joker, who reminds us of our dark humanity.

Who are your heroes in real life?

Anonymous people help others, but they do not appear in newspapers or social media.

What aphorism or motto best encapsulates your life philosophy?

"Stairs are climbed from the front, since climbing them from behind or the side will result particularly uncomfortable." — Julio Cortázar, "Instrucciones on How to Climb a Staircase" (from Cronopios and Famas, 1962). Trans. Paul Blackburn.

In the original: "Las escaleras se suben de frente, pues hacia atrás o de costado resultan particularmente incómodas." — Julio Cortázar, "Instrucciones para subir una escalera" (de Historias de cronopios y de famas, 1962).

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