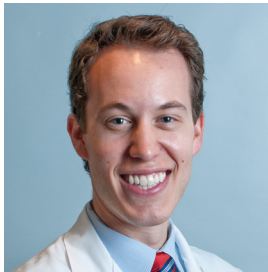


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Dr. Brian Skotko: Collaboration Leads to Better Outcomes for Children with Down syndrome



Dr. Brian Skotko

Working as a clinician and researcher has always been Dr. Brian Skotko's dream and in many ways, it all starts with his sister, Kristin. Kristin, who recently turned 40, has Down syndrome and has been Skotko's "life coach," he says.

Today's he's living that dream after studying pediatrics, genetics, and public policy, and serves as the Emma Campbell Endowed Chair on Down syndrome at Massachusetts General Hospital where he is director of the hospital's Down syndrome Program. In addition to numerous academic publications, he also co-authored nationally award-winning books, *Common Threads: Celebrating Life with Down Syndrome* and *Fasten Your Seatbelt: A Crash Course on Down Syndrome for Brothers and Sisters*.

Skotko says collaboration is especially crucial for the Down syndrome community because there are simply not enough resources nor time to unnecessarily compete. Because any single pediatric clinic serves only a handful of patients with Down syndrome, recruiting for clinical trials can take years.

But if researchers work together, they can find larger numbers of participants for studies and get better evidence for what works.

He cites a recent example: anemia. Iron deficiency is common in children with Down syndrome (just as in the general population) and can lead to cognitive declines. For children with Down syndrome who already have cognitive delays, anemia is a problem that urgently needs to be diagnosed and treated. Two years ago, researchers from around the world shared data on patients with Down syndrome and identified anemia as a significant concern for this population.

"I want the world to know that there has never been a better time to live with Down syndrome than now. Research has never been so vibrant. People with Down syndrome are participating in research and asking questions. Real answers are around the corner." Dr. Brian Skotko



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By working as a consortium, they had enough data to recommend new guidelines for how to better screen children with Down syndrome for iron deficiency. This collaboration changed the practice of medicine.

Skotko said this research was completely unfunded, but clinicians who care for people Down syndrome stretched themselves to make the consortium work. He is optimistic that these researchers might be able to go even further, if they secure one of the grants from INCLUDE funding.

“Clinicians are frustrated with not having evidence to help them guide families, but we know we have the potential to find even more answers with funding,” he said. “I want the world to know that there has never been a better time to live with Down syndrome than now. Research has never been so vibrant. People with Down syndrome are participating in research and asking questions. Real answers are around the corner.”

“We often work with families on recruitment and how to advertise studies, but it’s still novel to have them be actual co-designers of the entire research process. When we engage patients and their families in that process, it’s very informative and the outcome of the research is stronger.”

Dr. Brian Skotko

Learn more about how researchers have worked to improve screening for anemia in children with Down syndrome:

Hart, S.J., Zimmerman, K., Linardic, C.M. et al. Detection of iron deficiency in children with Down syndrome. *Genet Med* 22, 317–325 (2020). <https://doi.org/10.1038/s41436-019-0637-4>

His advice to researchers going forward is to view families as more than people to recruit but as actual collaborators. “We still have a lot more room for improvement in making sure to include families in designing research,” he said. “We often work with families on recruitment and how to advertise studies, but it’s still novel to have them be actual co-designers of the entire research process,” he said. “When we engage patients and their families in that process, it’s very informative and the outcome of the research is stronger.”

He also urged people with Down syndrome and their families to continue making sure their voices are heard. “Step up to the microphone to remind people why funding is important, remind them of the day-to-day questions. Our questions are always more relevant when informed by families,” he said.



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