OPINION

Uncommon testing

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Given how often I write about genetics and autism, I recently asked a friend who has a son with autism whether she had considered — or even been offered — genetic testing. Her reply is probably characteristic of that of many parents: “We thought about it but decided to focus on things we knew could help him,” such as **intensive behavioral therapy**.

A new study, published 14 March in the *Journal of Genetic Counseling,* confirms the **relative infrequency of genetic testing** for autism, despite the fact that three major professional societies, the American Academy of Neurology, the American Academy of Pediatrics and the National Society of Genetic Counselors, recommend that families of children with autism be offered **genetic testing and counseling.**

The clearest reason for genetic testing for autism is to inform parents who plan to have another child and want to better understand the risk of recurrence. Previous research shows that parents often over- or underestimate recurrence rates.

I asked someone on the front lines of genetic testing, **Wendy Chung**, assistant professor of pediatrics at Columbia University in New York and principal investigator for the **Simons Variation in Individuals** project (VIP), supported by SFARI.org’s parent organization, about her experience with genetic testing for autism. She says parents most likely to seek a specific diagnosis are “those considering having more children, those with children who have regressed especially, those concerned about prognosis, and a few wanting to ensure they have not overlooked any etiologies with specific treatment.”

Currently, only about 5 to 15 percent of cases of autism spectrum disorders have a detectable genetic cause (although that may soon change as genetic studies identify additional autism-linked mutations). And identifying a genetic cause of the disorder rarely informs treatment. But because

some genetically defined causes of autism come with risk of other health problems — deletions of 22q11.2, for example, can also be linked to cardiac, renal and immune issues — detecting such mutations can encourage screening and prevention.

In the new study, researchers surveyed 155 families, asking whether they'd had genetic testing or counseling and why or why not. Twenty percent had seen a genetics professional and about 30 percent had undergone genetic testing.

Those who did get testing were more likely to have had a doctor suggest it, or to know someone with a genetic cause of autism.

Physicians may be to blame for low testing rates: Among families who hadn't seen a genetics professional, about half said it was because their physician hadn't suggested it; and nearly 60 percent of those who got a referral to a geneticist had asked for it, implying "that providers might actually be less aware than is estimated by the referral data alone," the researchers write.

They say the results show that medical professionals and families need to be better educated on the existence and value of genetic testing for autism.

It turns out that a minority of parents share my friend's reasoning: Seventeen percent of those who hadn't seen a genetics professional said it was because they didn't see the value, mostly because they didn't think it would help their child's treatment.

"Although it's still not critical to know the molecular genetic type of autism (unless seizures are involved), I like to know for my patients so I know what clinical trials I need to monitor for each patient," says Chung.