Continuing the Debate on Autism and VCFS: What Does It Mean For Treatment?

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The report of Angkustsiri et al.[1] that was the basis of the preceding editorial extends beyond the world of VCFS to the way that we diagnose and assign the autism "label" in general. The percentages of children who have another genetic diagnosis, and yet are classified as "autistic", are not widely available despite the fact that frequency statistics for autism are reported and diffused each year. In point of fact, I searched for the overlap statistics for the purposes of this editorial, and could not find them. In the case of VCFS, up to 50% of cohorts have presented with autistic symptoms,[2] but as Angkustsiri and colleagues eloquently showed, very few of those children probably meet criteria for an autism spectrum disorder.

The main confusion stems from the fact that autism is diagnosed through the presence of symptoms, or namely, communication difficulties, social challenges, and the presence of repetitive behaviors (DSM-5). Of course, communication difficulties and social challenges are present in a large number of disorders, particularly neurodevelopmental syndromes with genetic or chromosomal mutations. As Angkustsiri et al stated,[1] children affected by VCFS often appear to meet autistic criteria on paper, but their social motivation and social interest is very different from a child on the autistic spectrum disorder. To understand this difference, one needs to spend time with the individual child.

It is, however, understandable that we have arrived at this crossroads. We are not far enough along in our understanding of the origins of autism to trace putative causes and be more specific in our diagnosis; however, grouping children on the basis of common traits is necessary and helpful for research and intervention. Early risk patterns of autism (assessed in infants using the *Autism Observation Scale for Infants*), have led to their inclusion in promising early intervention programs, such as the Early Start Denver Model.[3] However, it is not yet clear whether such interventions are effective exclusively for children with autism, or whether they would help a variety of children with developmental problems that include delays in communication and social skills.

Perhaps it does not matter. Researchers and clinicians alike continue to arrive at the same conclusion: educational and treatment programs need to be

individually tailored to the child, whether or not we understand the biological or symptomatic nature of his/her disorder. A valid short-term goal would be to have enough effective treatments in our arsenal that we can put together a tailored program for every child. Fortunately, treatments can be effective for children who have different underlying diagnoses. Angkustsiri et al mentioned a recent study of the computerized cognitive remediation program, *Vis-à-Vis*,[4] which was subsequently tested with three groups of children: those diagnosed with autism only, those diagnosed with VCFS, and a group with idiopathic developmental delay. To our surprise, all three groups improved after using *Vis-à-Vis*, however, their gains were in different domains, as were the localization of functional brain changes after using the program. Interventions and treatment programs may be relevant for different groups of children who share certain cognitive difficulties or phenotypic characteristics, but they must be chosen for that particular child.

There are excellent research groups who are working hard to understand the biological underpinnings of autism. Recent results from DNA methylation studies,[5] placental morphology,[6] postmortem neuronal work[7] and gene expression [8,9] offer exciting glimpses into knowing more about the root causes of autism. However, until we can make our "autistic groups" more biologically homogeneous, we need to be aware that the label "autistic" currently includes widely heterogeneous individuals, with heterogeneous social handicaps and, often, heterogeneous congenital disorders underlying their symptoms.

References

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