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Not all "autistic traits"are autistic, nor do all complex genetic conditions have a poor prognosis

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Statement of the Problem: Complex genetic phenotypes are a therapeutic challenge in autism. Earlier recognition/treatment associate with better prognosis but less diagnostic stability. A boy´s initial symptoms erroneously suggested autism. Methodology & Theoretical Orientation: Clinical case report; follow-up from 2y6m to 5y9m. Findings: The boy was born well by cesarian section at 35th week after continuous bleeding during pregnancy. He presented hypospadias, hypotonia, unresponsiveness, difficulties in feeding (no rooting reflex, nonacceptance of breast/bottle). Head circumference in 50th percentile. Weight remained below 3rd percentile. Gastrostomy feeding after 15m improved nutrition. EEG and NMR were normal. ArrayCGH demonstrated microdeletion (9p24.3p24.1) and microduplication (8q24.11q24.3), previously described in two cases. At 2y6m, he was unable to sustain neck and trunk, making scarce use of hands, reaching for objects with feet. Pain reactivity was low, hurting without crying. Sialorhea with impairment of orofacial tone/sensitivity were associated with food refusal, including small water volumes. He responded well to physical therapy with TREINI Exoflex exoskeleton, being able to stand and walk alone within 90 days. Response to speech therapy intervention was slower, but effective. The only articulated is "mamá" and he presents orofacial dyspraxia. Contextualized verbal comprehension is very good. He engages in pretend and sociodramatic play and is learning to use an AAC. Cognition and sociommunicative abilities improved from 10th to 25th percentil from ages 4y to 5y. If present, intellectual disability is mild. Following diagnoses were formulated: a) syndromic: hypotonic cerebral palsy; Worster-Drought syndrome; speech/limb apraxia; b) topographic: perisylvian dysfunction; c) Etiological: microdeletion (9p24.3p24.1) and microduplication (8q24.11q24.3); d) functional: see Figure 1. Conclusion & Significance: Traits such as food selectivity/refusal, lack of social and painful reactivity is present in conditions other than autism. Children with and complex genetic conditions may respond well to intervention.

Biography

Vitor Geraldi Haase is a physician with medical residency in neurology and pediatric neurology, PhD in medical psychology, retired full professor of developmental neuropsychology, currently working as consultant at TREINITEC, a private company developing intervention programs for children and adolescent with neurodevelopmental disorders. Mai research interestes are related to motor impairments underlying neurodevelopmental disorders.

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