A case study of a 21-year-old adult with Angelman syndrome with fluent verbal and independent functional living skills.

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The Angelman syndrome (AS) Natural History Study is a longitudinal observational study that aims to assess development, behavior, and communication in a large group of individuals with AS. We will present the case of an individual with AS seen in the AS Natural History Study who demonstrates fluent verbal communication skills and a high level of independent functional living skills. This individual (subject A) is a 21-year old woman with AS due to abnormal methylation of the SNRPN gene. Subject A has experienced some characteristic symptoms of AS, including difficulties in feeding during infancy, sleep problems, and global developmental delay. However, her presentation is relatively mild compared to most adults with AS. She currently attends a boarding school and is employed through school at a child care facility.

An in-depth intake interview was conducted to obtain an overview of subject A’s medical and developmental history and current functioning. In addition, cognitive functioning (verbal and non-verbal) and adaptive functioning were assessed using the Leiter International Performance Scale, the Vineland Adaptive Behavior Scale-Third Edition (Vineland-III), and the Wechsler Abbreviated Scale of Intelligence (WASI-II). Relevant information from the interview, behavioral observations, and standard scores and age equivalents from the assessments will be presented. Subject A demonstrates higher adaptive functioning and self-help skills, verbal conversational skills, and motor skills than are typically seen in individuals with AS. Her standardized Vineland-III age equivalents range from 3-15 years across the communication, daily living, and socialization domains. Subject A’s adaptive functioning skills will also be compared to other AS adults using data from the Natural History study. This case demonstrates that AS should not be ruled out as a diagnosis in individuals with verbal communication and high adaptive functioning skills. Additionally, this case provides evidence that there is more diversity in verbal ability and functional living skills in AS than has been previously reported in scientific literature.