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Short Communication

Genetic Sequencing and Understanding Autism Spectrum Disorder (ASD) Deeper at a Genetic Level

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INTRODUCTION

Autism spectrum disorder (ASD) is a term that refers to a group of early-onset social communication deficiencies and repetitive sensory–motor activities that can be caused by a significant genetic component as well as environmental factors.

Autism Spectrum Disorder (ASD) is a spectrum of complex neurodevelopmental diseases marked by repetitive and characteristic patterns of behavior as well as social communication and interaction impairments. The symptoms appear in early childhood and interfere with daily activities [1].

The term "spectrum" means the vast range of symptoms, skills, and levels of functional handicap that people with ASD might experience. Some children and people with ASD are entirely capable of doing all daily activities, while others require significant assistance to do so [2].

Pervasive Developmental Disorders are a category of childhood mental illnesses in which social skills, communication, and behavior do not develop properly or deteriorate during development. These disorders are relatively common, affect various mental functions, and cause a degree of chronic and persistent disability. Early diagnosis and treatment reduce the morbidity associated with these pathologies. Previously referred to as early-onset psychosis, patients with autistic disorder present severe difficulties in communication, social isolation, mannerisms, and stereotyped behaviors, as well as attraction to inanimate objects, great mental rigidity, and even obsessive behaviors [3,4].

PTEN Hamartoma Tumor Syndrome (PHTS), Fragile X, RETT Syndrome, Phelan McDermid Syndrome, and others are all linked to autism spectrum disorder (ASD).

Because of the widely diverse observable somatic abnormalities representation seen on patients1-2, clinical diagnosis of Autism Spectrum Disorder has been difficult. Because there was limited molecular understanding of ASD in the beginning, traditional clinical diagnosis of ASD was primarily focused on syndromes. With multidisciplinary interview-based examinations comprising speech and language therapists, paediatricians, child development centre psychiatrists, and psychologists1-3, these symptom-based diagnostic tests could be time-consuming. Many ASD patients with complicated ASD symptoms do not receive an accurate diagnosis until later in life, depriving them of the opportunity to receive appropriate therapy [5].

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