

The Role of Primary Healthcare Providers in Early Screening and Identification of Markers for Rare Comorbid Manifestations of Autism

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ABSTRACT

Although Autism itself is not considered a rare neurodevelopmental disorder, it is closely linked with several neurodevelopmental rare diseases. Early surveillance, detection, screening, and referral pathways are essential steps to lead to and inform an evidence-based diagnosis of autism spectrum disorders. The role of primary healthcare providers in early screening and identification of early signs of autism spectrum disorder, phenotypes, and behavioral symptoms is essential and instrumental in early identification of comorbid rare disease and access to early medical care. Exploring the role of primary healthcare providers in identifying markers for rare comorbid manifestations of autism in primary healthcare is a systematic process to quickly identify any early indicators of autism spectrum disorder and identifying markers for rare comorbid manifestations of autism in primary healthcare. It is an integral pathway to inform and facilitate a cohesive and comprehensive multi-disciplinary diagnostic protocol. The attendees will be introduced to the targeted functions of standardised screening questionnaires, parental interview forms and checklists covering a broad indication of issues relevant to general behavior, communication, sensory issues, ASD and/or social communication difficulties, any special interests and developmental, as well as an indication of whether more in depth diagnostic assessment is advisable for further investigations of comorbid genetic and /or rare diseases. Finally, we will look at how to draw and use essential preliminary information that will contribute to an initial clinical impression indicating the need for further diagnostic assessments, and unlocking in-depth assessment and specialized medical and behavioural care pathways.