Updated clinical practice recommendations for managing children with 22q11.2 deletion syndrome


This set of clinical practice guidelines focuses on the care of individuals from birth to 18 years of age. The emphasis is on periodic assessments and family-centered care.

Early diagnosis and treatment as well as preventive management are strongly recommended as they optimize health, functioning, and quality of life. Healthcare providers need to have basic knowledge about the variable, multi-system, and changing nature of 22q11.2DS. Specialists from various disciplines need to provide coordinated care and tailor the treatment to suit the child. Family members and caregivers are an essential part of the care team and benefit from information and support.