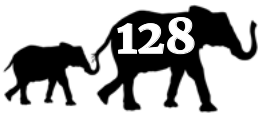

128: Patients with 22q11.2 deletion syndrome - Too challenging to treat?

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Background: The complexity of individuals with 22q11.2 deletion syndrome (22q11.2DS) may be daunting to many health care professionals, and to caregivers. This may especially be the case with respect to the psychiatric features of this genetic condition. **Methods:** Using a narrative (qualitative) study method, we reflected on the wisdom of clinicians with over 100 collective years of experience with 500+ adult patients with 22q11.2DS at a single dedicated centre. **Results:** While undoubtedly recognizing the multi-system nature of the genetic condition, most clinical specialists and general practitioners accepted the challenge of caring for patients with 22q11.2DS. Disproportionately, however, psychiatric specialists and clinical teams in the community were exceptions in this regard. Many early psychosis intervention programs routinely exclude patients with developmental delay and/or intellectual disability, and many psychiatrists turn away patients with 22q11.2DS patients, citing complexity. Attributing signs and symptoms of serious but treatable psychiatric illness to "behaviour" often led to years of multiple therapy trials before accurate diagnosis and implementation of standard treatment. From the family/caregiver perspective, fears of psychiatric diagnostic labels and/or of evidence-based medications hindered or delayed delivery of standard-of-care, in contrast to usually immediate acceptance of comparable neurologic, endocrinologic, or cardiac diagnoses and treatment recommendations. Help-seeking in alternatives such as non-evidence-based therapies, or self-treatment with actively harmful alternatives such as cannabis, was common. **Conclusions:** If individuals with 22q11.2DS were known to be at a 1 in 4 risk of developing a severe yet treatable form of non-psychiatric illness (e.g., cancer), optimizing diagnosis and providing standard-of-care management would likely be a matter of urgency to healthcare systems, clinicians, and caregivers. Mechanisms to address systemic issues in clinical practice, and in the general population, related to recognizing psychotic illness and considering individuals with serious but treatable psychiatric illness worthy of care, are needed.