Delayed Diagnosis and Intervention of 22q11 Deletion Syndrome in a 17-Year-Old Male with Psychosis: a Case Report

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Background

What is Human 22q11 Deletion Syndrome?

- Prevalence: Occurs in about 1 in 4,000 births.
- Presentation: Includes heart defects (e.g., tetralogy of Fallot), immune deficiencies, cleft palate, and distinct facial features.
- Diagnosis: Detected through genetic testing such as FISH.
- Management: Involves surgical interventions, immunotherapy, developmental therapies, and psychological support.

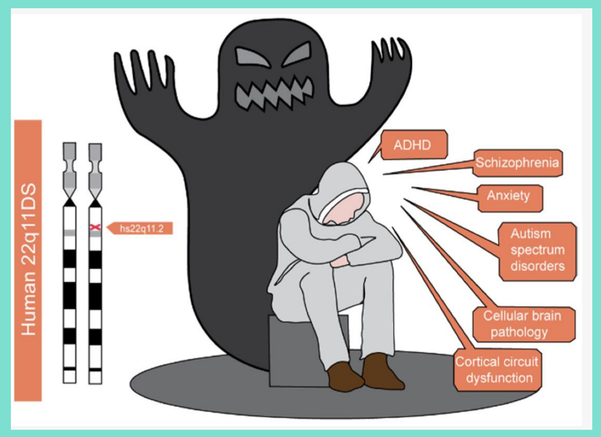

Figure 1: de Oliveira Figueiredo EC, Bondiolotti BM, Laugeray A, Bezzi P. Synaptic Plasticity Dysfunctions in the Pathophysiology of 22q11 Deletion Syndrome: Is There a Role for Astrocytes? International Journal of Molecular Sciences. 2022; 23(8):4412. https://doi.org/10.3390/ijms23084412

Cognitive effects of 22q11 Deletion Syndrome:

- Impaired cognitive functioning
- Decreased attention and focus
- Lower IQ than in individuals without deletion
- Higher incidence of psychotic disorders

IMPORTANT TO NOTE: Psychotic disorders co-morbid with 22q11 deletion syndrome are more treatment resistant

Case Description

Patient Background:

- Age/Gender: 17-year-old male
- Language: Mandarin-speaking
- PMH: VSD, RBBB, hypothyroidism
- PPH: Psychotic symptoms starting at age 15, no prior diagnosis

Inpatient admission risperidone/valproic acid- no improvement → Inpatient admission chlorpromazine/ sertraline- no improvement → Inpatient admission- clozapine- some improvement in psychosis

Current presentation:

- new onset aggressive behavior towards father
- auditory hallucinations, paranoia,
- grandiose delusions (divine identity)
- bizarre delusions (possession by an unidentified entity)
- magical thinking

Abnormal labs: TSH of 7.68 Utox was negative.
MSE: significant for abnormal facies, pacing, thought blocking, appearing internally pre-occupied, playing cards with an invisible partner

Differential diagnosis:

- Primary Psychotic Disorder Decompensation: Ruled out due to adherent clozapine treatment.
- Medical Causes: Ruled out due to stable thyroid levels post-treatment, negative UTox
- Genetic Condition: High suspicion, confirmed by genetic testing for 22q11 deletion syndrome, given history of ventricular septal defect, hypothyroidism, and abnormal facies.

- **Genetic testing revealed a diagnosis of 22q11 deletion syndrome**

- Family psycho-education was provided by a Mandarin speaking psychiatrist
- Referrals for cardiology and endocrinology
- Unfortunately lost to follow-up in our system

Discussion

- Given the high prevalence of psychiatric co-morbidities in patients with 22q11.2 deletion syndrome, we recommend considering genetic testing for cases presenting with other potential components such as ID, VSD, and abnormal facies.
- Limited English proficiency and understanding of the healthcare system can lead to delays in diagnosis and care
- Psychiatrists have a mandate to practice with cultural humility.

- The diagnosis and management of psychosis secondary to 22q11.2 deletion syndrome requires a comprehensive clinical approach including a multi-specialty team.
- Early diagnosis and intervention can improve medical and psychiatric outcomes.

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