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The Differential Diagnosis of Congenital Disorders That Include Psychosis

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BACKGROUND: Neuropsychiatrists are often called upon to evaluate psychotic individuals for possible neurological or neurodevelopmental etiologies after acquired neurological and other medical disorders have been ruled out. A large number of relatively rare congenital neurodevelopmental conditions that include psychosis have been described. Clear guidance on the neuropsychiatric evaluation and differential diagnosis of these conditions is difficult to find in standard textbooks.

OBJECTIVE: To address this dearth of information we set out to concisely describe the neurodevelopmental disorders in the differential diagnosis of psychosis, their neurodiagnostic and laboratory evaluations, and relative prevalence.

METHODS: A literature search was conducted for disorders that may present with psychosis, utilizing PubMed and Ovid, with search terms including psychosis, metabolic, genetic, congenital and neurodevelopmental disorders. All disorders described in case reports or case series and literature reviews, including their references, were initially included. Epidemiological and diagnostic information was gathered via textbooks, OMIM, GENETests®, and orphanet®.

Exclusion Criteria: 1. Acquired (non-heritable/non-congenital) disorders
2. Fewer than 3 cases reported with psychosis
3. Poorly described psychosis

Analysis: Disorders were categorized as follows:
1. By the presence of one or more of 20 associated signs (Table One)
2. Disorders having major associated signs are presented in Tables Two and Three along with principal diagnostic tests
3. By prevalence (> 1/10,000; 1/10,000-1/50,000; <1/50,000)

RESULTS: We identified 61 congenital disorders that may present from childhood through middle age and include psychosis.

• 44 disorders (72%) have prominent associated neurological features that facilitate differential diagnosis.
• 17 disorders have readily recognizable unique phenotypes.
• 44 disorders may present without mental retardation.
• 52 disorders (85%) have characteristic laboratory features.
• 52 have known loci and 3 disorders have loci yet unknown.
• 5 disorders were due to chromosomal nondisjunction.

DISCUSSION:
1. Case-report based research such as this is limited by difficulty in determining whether a reported relationship is coincidental or causal.
2. The cost of doing an exhaustive laboratory evaluation of all possible disorders that could result in psychosis would be astronomical. A coherent neuropsychiatric approach, such as the one presented here, increases cost savings by providing a probability-guided, examination-based approach to focus the workup.
3. Accurate neuropsychiatric diagnosis guides genetic counseling and treatment planning.
4. Studying neuropsychiatric disorders of known etiology that include psychosis will ultimately lead to research aimed at understanding the etiology of psychotic symptoms in Axis I disorders.

CONCLUSION: As consultants frequently called upon to evaluate atypical presentations of psychosis, neuropsychiatrists should be aware of congenital disorders that can present with psychosis, however rarely.

We recommend a differential diagnostic approach based on estimated prevalence of the disorders and their most prominent associated neurological features.

REFERENCES:
2. GENETests: http://www.genetests.org
3. Orphanet: http://www.orpha.net

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