**22q11.2 deletion and increased risk of schizophrenia in children and adolescents**

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**Objective:**
22q11.2 deletion [del.22q11.2] is the most frequent microdeletion in humans with an estimated occurrence of approximately 1 in 4,000 life births. Patients can show a variety of somatic symptoms, especially facial dysmorphia, congenital heart defects, thymic hypo- or aplasia, and velopharyngeal dysfunction with or without cleft palate. Developmental delays of psychomotor functions and language are very common, and about 40-50% of the affected individuals are mentally retarded (IQ < 70). Furthermore, the deletion seems to be one of the most important risk factors for schizophrenia. Until now, increased prevalence rates of this disorder have been well documented for adults, but not for children and adolescents with del.22q11.2 (1). Therefore, the aim of this study was to investigate whether there is an increased risk of schizophrenia for this age group.

**Method:**
Probands:
With support of the German 22q11.2 deletion syndrome foundation, KiDS:22q11, the primary caregivers of all known subjects aged 8-17 years (n=49) were anonymously asked to fill out questionnaires. The primary caregiver of another subject with del.22q11.2 could be recruited by an outpatient clinic.

Study design:

**I. Screening for schizophrenia:**

- Special questionnaire to compile personal, somatic and psychosocial history
- Fear of a physician or a primary caregiver that the proband could suffer from schizophrenia
- Pre-existant diagnosis of schizophrenia
- Child Behavior Checklist (CBCL) 4-18 (2)
- Hints for hallucinations (questions 40 & 70)
- Hints for bizarre behaviour or ideas (questions 84 & 85)

**II. Personal examination**

Case report:
Paranoid schizophrenia; age of onset: 12 years; diagnosis of del.22q11.2 at the age of 14 years. No congenital heart defect, but typical facial signs of the deletion, and hypocalcemia.

CBCL 4-18 (2):
Significantly higher t-values could be found for these two probands when compared to the other subjects of this study for the subscales "withdrawn" (p<0.005), and "thought problems" (p<0.010).

**Results:**

**I. Screening for schizophrenia:**

- 14 year old boy with known schizophrenia
- 14 year old boy with known schizophrenia
- Pre-existant diagnosis of schizophrenia
- CBCL 4-18 (2):
  - Significantly higher t-values could be found for these two probands when compared to the other subjects of this study for the subscales "withdrawn" (p<0.005), and "thought problems" (p<0.010).

**II. Personal examination**

Only the 14 year old boy with already diagnosed paranoid schizophrenia could be examined personally. According to CASCAP-D (3) he showed: hyperexcitability; generalized anxiety; persisting optic hallucinations; and cognitive deficits despite of neuroleptic medication (quetiapine). Standardized testing (e.g. intelligence tests) could not be done because of the severity of his psychiatric disorder. He met ICD-10 criteria for a schizophrenic residuum.

**Conclusions:**
About 10% of schizophrenic disorders seem to have an onset before the age of 17 (4). Taking this into account, according to our results, the prevalence of schizophrenia among children and adolescents with del.22q11.2 could be 20 times as high as in the general age group. There are some limitations of this study: quite low response rate, missing representativeness, lack of objective informations about intellectual capacities of participants, and impossibility of personal examination of the second adolescent with a possible schizophrenic disorder. Nevertheless, the results of this study indicate that children and adolescents with del.22q11.2 should be carefully screened for symptoms of schizophrenia at regular intervals.

**References:**

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