P02-167 - **COGNITIVE ALEXITHYMIA IN AN ADULT FEMALE WITH RING CHROMOSOME 21 SYNDROME (R(21))**

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A 30-year-old female patient with moderate mental retardation and minor dysmorphisms was referred for neuropsychiatric examination because of psychotic and autistic symptoms and impulsivity. She was born dysmaturally after an uncomplicated pregnancy. Postnatally, she developed cyanosis, feeding problems and recurrent urinary tract infections. Her developmental trajectory showed delayed milestones, speech retardation and restricted peer interactions resulting in special education from age 9. Behavioural problems because of sensory impairments induced paranoid ideation due to social misinterpretations, increasing from age 18.

At referral, physical investigation revealed mild dysmorphisms, severe hearing loss and visual impairment, oestrogen deficiency, kyphoscoliosis, mild hypotonia and hyperlaxity of joints. Neuropsychological examination disclosed a disharmonic intellectual and social cognitive profile (total IQ: 50) with adequate performal capacities and marked cognitive alexithymia. No formal psychiatric diagnosis could be established. Karyotyping showed a de novo ring chromosome 21: 46,XX,der(21)r(21)(p11q22.3). High resolution array analysis of chromosome 21 demonstrated one interstitial duplication, two interstitial deletions and one terminal deletion.

Ring chromosome 21 syndrome is a rare condition that may occur on a de novo basis or via parental transmission. Major characteristics of the phenotype are vulnerability to infections, mild facial dysmorphisms, infertility and kyphosis. In addition there may be congenital malformations of nearly all organ systems.

The current case report demonstrates the importance of targeted high resolution micro array analysis in order to substantiate the genotype-phenotype correlation in patients with r(21) and stresses the importance of cognitive alexithymia as a potential cause for behavioural problems and psychiatric symptoms in patients with mental retardation.

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