Background and Aims

DiGeorge syndrome (DGS) is a rare disease associated with a microdeletion of chromosome 22q11.2. Among clinical signs: heart defects, immunological alterations, psychiatric disorders. DGS children present developmental delay. The aim of this study is to assess cognitive and behavioural development of DGS paediatric patients.

Methods

Cognitive profile was assessed in a prospective cohort of DGS children referred to Paediatrics Department- Padua University (1998–2012). For a sample of 20 children (11 females, 9 males; 25% < 2 yrs, 25% 3–5 yrs, 50% > 6 yrs), informations were collected on diagnosis, surgical interventions, hospitalizations, treatments/rehab training programs. Cognitive profile was assessed using Griffith’s Mental Development Scales (GMDS) and Wechsler Intelligence Scale for Children-III (WISC-III), depending on children’s age. Behavioural profile was assessed using Child Behavior Checklist (CBCL). Univariate and multivariate descriptive analyses were performed.

Results

For younger children (GMDS, 10 children), global mental development resulted: 15.4% moderate retardation, 61.5% mild retardation, 30.8% borderline, 7.7% low normal, 15.4% normal. Worse scores are observed in the subscales: language, performance, eye-hand coordination and practical reasoning. For older children (WISC-III, 10 children), 76.9% had Mental Retardation (15.4% moderate MR, 61.5% mild MR), and 23.1% got low Global IQ scores (7.7% borderline, 15.4% low normal). Behavioural profile is barely normal in pre-school children and becomes borderline/clinical in school children (100% disadaptive functioning, 40% internalizing problems, 20% externalizing problems).

Conclusions

DGS patients have a wide spectrum of developmental delays, which require tailor-made rehab programs, and a worsening in behavioural profile in pre-adolescence and adolescence.