The clinical, cognitive and motor features in CHARGE syndrome: case study and new tools for diagnostic, therapeutic and rehabilitative management

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INTRODUCTION

CHARGE Syndrome is an autosomal dominant genetic disorder typically caused by mutations in the Chromodomain helicase DNA-binding protein 7 (CHD7) gene [1]. The incidence is estimated at 1/8,500–12,000 births [2]. The term CHARGE is an acronym that denotes the six major clinical features: ocular Coloboma, Heart defects, Atresia of choanae, Retardation of growth and/or development, Genital and/or urinary anomalies, Ear anomalies [3].

CHARGE children are affected by plurisensorial disabilities and therefore they have many medical problems not evaluable by already-known developmental scale.

OBJECTIVES

The purpose of our study is to edit psychometric tests and questionnaires for evaluation of various abilities of patient with CHARGE syndrome, to improve diagnostic, therapeutic and rehabilitative management of patient with CHARGE syndrome, to delineate the natural history of the disease more thoroughly and further to study and define patients’ cognitive and motor disabilities.

MATERIALS & METHODS

The Pediatric Clinic of the Institute of Maternal-Infantile Sciences in Ancona, in collaboration with the Lega del Filo d’Oro in Osimo (An), a non-profit organization for people with multi-sensorial impairments, take care of approximately 60 patients with clinical diagnosis of CHARGE syndrome, (in accordance with Blake’s classification [4]). Patients aged between 0 and 35, from all over Italy. Because of the very big number and the very wide geographical distribution of our patients, our case can be representative of the Italian population of CHARGE syndrome.

Each patient was subjected to a diagnostic and rehabilitative procedure including specialist assessments and laboratory and instrumental examinations. They were also evaluated for normal or delayed development in cognitive and motor abilities using an Italian questionnaire “Guida ai progressi del bambino” (Progress Guide). The questionnaire was composed of 10 domains: gross-motor skills, fine-motor skills, cognitive skills, socialization and play, self-care (feeding, washing, dressing, sphincter toileting), communication and expressive skills.

The level of abilities reached by each subject was assigned by a trained professional and expressed as age-equivalent score. The genetic survey on the CHD7 gene was carried out by an MPLA test – Ligation Probe-dependent Amplification.

REFERENCES


RESULTS

Data obtained from the diagnostic procedure showed a multi-systemic involvement and provided an overall assessment of each individual. This enabled an early rehabilitative and therapeutic approach. From the Progress Guide it was possible to investigate totally and partial skills and also to extrapolate a graph in which acquired skills at different times of patients’ follow-up are compared. Within the rehabilitation course this enabled to implement a personalized interventions.

CONCLUSIONS

The “Progress Guide” is a tool that can identify, in a simple manner and with a certain flexibility, the behavioural characteristics of multi-disabled children by comparing them with non-disabled children. The tool also makes it possible to identify the areas of strength and weakness of any subject and to monitor the development that takes place during psychoeducational interventions.

Statistical analysis has showed that our patients have delayed development in all skills with respect to chronological age. In particular, the delay is statistically significant in terms of self-care skills (worse toileting, better washing) and the communication skill (language); on the other hand, the expression skill is still preserved.