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Psychomotor development in Cri du Chat syndrome: comparison in two Italian cohorts with different rehabilitation methods.

The Cri du Chat syndrome (CdC) is a rare genetic disorder caused by variable size deletions of the short arm of chromosome 5 (5p-).

It is well known that home – reared patients show better performances as compared to institutionalized cases, and it was reported that continuous educational intervention can ameliorate their performances.

To asses the efficacy of educational intervention and to develop new CdC oriented programs of rehabilitation, we compare obtained results in two groups of CdC patients undergoing two different rehabilitation programs for many developmental skills.

We used data on the development of a group of CdC patients obtained by validated Italian translation for the Denver Developmental Screening Test II. We compared a group of 13 patients undergoing an educational program developed for CdC patients, the Mayer Project (MP), to a second group of 15 patients whose care was not specifically oriented. All patients were born in 2005 or later. A.B.C. (Associazione Italiana Bambini Cri du Chat) offered their families the possibility to be followed by the same expert in special education (ED) who suggested the type of educational intervention. ED offered to contact the families twice a year. ED then provided personalized and specific suggestions including sensory stimuli and motor opportunity to improve the ongoing family and local professional care. This educational program is from now on referred as Mayer Project (MP).

A positive impact of the MP was reported by parents, observing an improvement in social skills obtained, even if no significant differences were observed when the items of the Denver test were studied. The need for personalized care in CdC patient and the choice of different methods to compare the result are also discussed.

The comparison failed to demonstrate significant differences between the two groups. However parents consistently reported improvement in everyday life. To evaluate any improvement test based on psychomotor development, which was intended to describe the neurological impairment, is not the best choice, and this is the explanation for the likely lack of significance. The educational program "MP" is still ongoing and collection of data based on different scales, to obtain a quantitative evaluation of parental observations about improvements obtained by the MP program are in progress.

		MAYER PROJECT	NON MAYER PROJECT
NUMBER OF PATIENTS		13	15
M/F		9M 4F	7M 8F
MEAN AGE AT DATA COLLECTION, YEARS (RANGE)		6.5 (3-9)	7 (2-9)
GESTATIONAL AGE AT BIRTH, WEEKS (RANGE)		36.83 (32-40)	37.13 (28-40)
ASSOCIATED MALFORMATIONS		-	-
HEAD CIRCUMFERENCE AT TERM OR CORRECTED AS TO 40 WEEKS GESTATIONAL AGE.		34	31.5
STANDARD KARYOTYPE: 5P-	BREAK POINT IN BAND		
	15.2	1	2
	15.1	2	-
	14	8	7
	13	1	1
	INTERSTITIAL DELETION	1	3
	UNBALANCED TRANSLOCATION	-	2

Table 1: Demographic and Cytogenetic features of the two groups of CdCS patients.

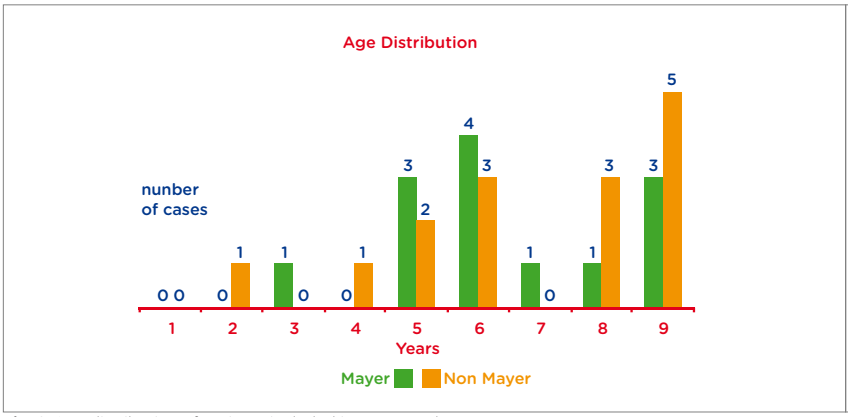

Fig. 1: Age distribution of patients included in Mayer and Non-Mayer group.

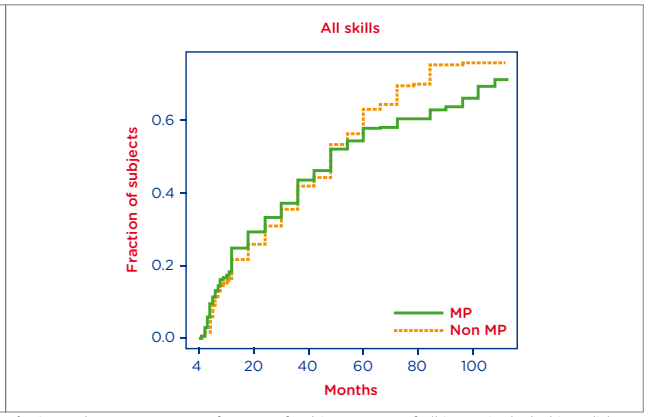

Fig.2: Kaplan Meyer curves for age of achievements of all items included in validated Italian translation for the Denver Developmental Screening Test I.