Life Span and Disability / XII, 1 (2009), 41-52

Genetic Syndromes Causing Mental Retardation: deficit and surplus in school performance and social adaptability compared to cognitive functioning

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Abstract

In this paper we reported some results of research carried out in Italy with participants with Mental Retardation (better defined as Intellectual Developmental Disability) due to genetic syndromes (Down, Fragile-X, Cornelia de Lange and Prader-Willi), evidencing specific conditions characterized by deficit or 'surplus' in reading, writing and maths performances, and in social adaptation respect to the intellectual competencies.
In some cases the comparison was made also with respect to abilities of memory and language.
Results suggested that the cases of 'surplus' are in our context more frequent than those found in International literature, and this may be due to the positive effects of the inclusion in mainstreaming classrooms of most pupils with intellectual disabilities.
A debate on these issues, comparing different cultural and social realities, is welcome.

Keywords: Intellectual Disabilities, Deficit, Surplus, Genetic Syndromes

Introduction

The progressive shifting from the expression 'Mental Retardation' toward the more adequate 'Intellectual Developmental Disabilities' was motivated by its even more frequent use in scientific literature and the presence of this expression in the name of different Associations (Luckasson & Reeve, 2001; Shalock, Luckasson, & Shogren, 2007).

* *Received*: 27 February 2009, *Revised*: 20 May 2009, *Accepted*: 20 May 2009.

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Moreover, the use of the term intellectual disability shows the shift of the researchers' interest from the study of mental retardation in general, to the study of the specific profiles of many syndromes causing mental retardation, better defined as Intellectual Developmental Disabilities (Dykens, Hodapp, & Finucane, 2000).

Comparison between Down syndrome and Williams syndrome cognitive profiles shows complementary strength and weakness, as regards verbal competencies (higher in Williams syndrome) and visuo-spatial skills (more efficient in Down syndrome) (Vicari, Carlesimo, Brizzolara, & Pezzini, 1996; Dykens *et al.*, 2000; O'Brien & Yule, 2000; Vicari, Marotta, & Carlesimo, 2004).

Besides research evidencing what the different syndromes have in common – e.g., deficits related to the "Central Executive" function (Lanfranchi, Cornoldi, & Vianello, 2004; Lanfranchi & Vianello, 2004; Lanfranchi & Vianello, 2006) – many other studies aim to search for specificity of syndromes cognitive aspects.

We will focus our attention on results of research regarding the cognitive-behavioral and adaptive profiles (including school performances) of four syndromes: Down, Fragile-X, Cornelia de Lange and Prader-Willi syndromes.

Down syndrome is the most studied, and the most frequent genetic syndrome. Different authors have underlined the specificity of the behavioral and adaptive profile: compared with the intellectual level, linguistic performances are lower, except for pragmatic aspects (Buckley, 1999; Vinter, 2002; Rondal, 2004a; Rondal, 2004b); deficits are found in short-term verbal memory and high-controlled working memory (Lanfranchi *et al.*, 2004); while visuo-spatial short-term memory are coherent with intellectual level, and adaptation skills are superior if compared with general intelligence (Vianello, 2006).

As regards the Fragile-X syndrome (the most frequent hereditary syndrome causing intellectual disability), literature has shown marked differences between genders, deficit in working memory and sequential memory, good simultaneous processing and adult adaptation (Saunders, 2000; Lanfranchi, Cornoldi, Drigo, & Vianello, 2008).

A different profile is shown in Prader-Willi syndrome (well-known for the hyperfagia), characterized by preserved abilities in the visual-motor discrimination if compared to auditory-verbal one, in the visual more than auditory attention, in the integration of spatial more than verbal stimuli, in simultaneous more than sequential processing, in long term more than short term memory (Waters, 1999; Dykens *et al.*, 2000).

A peculiar profile was found also in Cornelia De Lange syndrome, with particular points of strength, in visuo-spatial memory, perceptual organization and fine-motor behavior, and weakness in attention and language skills (Fiori, Lanfranchi, Moalli, & Vianello, 2008).

In most research an assumption is implicit, that inter-syndromes and intra-syndromes differences are mainly due to genetic differences.

We will suggest a line of research complementary to this, evidencing the interaction between biological bases and environmental influence, i.e. poor or normal or enriched environment (according to Baroff, 1989), producing cognitive and behavioral profiles of persons with genetic syndromes

The constructs involved are those of 'deficit' or 'surplus' with respect to mental age.

The 'deficit/surplus' hypothesis.

Education and learning can significantly influence academic and social abilities in individuals intellectual developmental disabilities. Applied research (Baroff, 1989, Vianello, 2008) has revealed the existence of two opposed, contrasting phenomena.

Zigler and Bennet-Gates (1999), on the basis of research by Zigler and co-workers over a period of 40 years, found that individuals with mental retardation show, at a motivational and personality level more negative behavior in the presence of strangers, psychological dependence on adult figures personally-known to subjects, less expectation of success, more importance given to external motivation rather than internal.

As a result of this, the person is less motivated to work, has less self-esteem, and self-efficacy. A consequence of this is the individual's taking less advantage of personal potential, and is therefore in "deficit" respect to mental age (Vianello, 2008); that is, performances below expectations of cognitive functioning. The phenomenon opposite to mental age deficit is characterized as "surplus". This issue has had very little literature dedicated to its existence. It analyzes how adequate educational intervention can facilitate above-average performance compared to typically developing children wuth the same mental age (or equivalent intellectual age resulting from intelligence tests).

In this paper we summarize the results of some studies conducted in Italy in samples of persons with mental retardation due to the genetic syndrome (Down syndrome, Fragile X syndrome, Cornelia de Lange syndrome, and Prader-Willi syndrome), showing the typical situations characterized by "deficit" or "surplus" in reading, writing, maths, and social adaptation compared to their assessed intellectual skills.

We specifically evaluated the variables: intelligence, assessed with the Wechsler Scale (Wechsler, 1974) or other measures less sensitive to educational influences, as *Logical Operation* Test (a simplified version of *Logical and Conservation Operations* test, Vianello & Marin, 1997) and "*Correspondence and Function Assessment*" (CFV, Vianello & Marin, 1998); adaptation (assessed by Vineland Scales); in some cases short-term memory and working memory, linguistic production and comprehension.

Deficit and "surplus" compared to Mental Age in Down Syndrome.

Vianello, Lanfranchi, Moalli, Petrillo and Sestili inproving an earlier study conducted on ten participants (Sestili, Moalli, & Vianello, 2006) have analyzed the relation between intellectual level and academic abilities (reading, comprehension, writing and calculation abilities) respect to a group of 19 persons with Down Syndrome.

Table 1 - *Mental age assessed with LO test, and academic performance of 19 individuals with Down Syndrome (age 13-14), primary and middle school*[2]

	Mental age	*Reading: Instrumental*	*Reading: Comprehension*	*Writing*	*Ability To Calculate*	
1	4.3	Can't read	Below 1st grade	Can't write	Below 1st grade	=
2	4.3	Can't read	Below 1st grade	Can't write	Below 1st grade	=
3	4.3	Can't read	Below 1st grade	Can't write	Below 1st grade	=
4	4.3	Can't read	Below 1st grade	Can't write	Below 1st grade	=
5	4.9	4th grade	3rd grade	2nd grade	End 1st grade	++
6	5.0	Can't read	Below 1st grade	Can't write	Below 1st grade	=
7	5.0	Beginning 2nd grade	1st grade	Beginning 1st grade	Below 1st grade	+
8	5.3	End 1st grade	Beginning 1st grade	Beginning 1st grade	Below 1st grade	+
9	5.6	1st grade	1st grade	Beginning 1st grade	Below 1st grade	+
10	5.6	1st grade	1st grade	Beginning 1st grade	Below 1st grade	+
11	5.9	Can's read	Below 1st grade	Can't write	1st grade, Interm.	=
12	6.9	Can't read	Below 1st grade	Can't write	Below 1st grade	-
13	7.0	2nd grade	4th grade	1st grade	End 1st grade	+
14	7.3	Beginning 1st grade	1st grade	Beginning 1st grade	1st grade, Interm.	=
15	7.3	End 1st grade	1st grade	1st grade	End 2nd grade	=
16	7.6	Beginning 1st grade	1st grade	Beginning 1st grade.	1st grade, Interm	-
17	7.6	End 1st grade	End 1st grade	End 1st grade	End 1st grade	-
18	7.6	5th grade	4th grade	2nd grade	End 1st grade	+
19	8.0	End 1st grade middle school	1st grade middle school	5th grade	3rd grade	+

As reported in Table 1 three children with Down Syndrome show a deficit (see last column with one less mark) in academic abilities respect to Mental Age, assessed in terms of "logical thinking" through the *Logical Operation* (LO) test, while eight children show a "surplus" (+ sign) more in reading, less in writing, and the least or completely absent in maths. It is our belief that this result reflects a different role of "logical thinking" in the three academic activities.

[2] We refer to Italian scholastic system.

Table 2 - *Intellectual level (Test of Logical Thinking), daily activities and socialization ability (Vineland test), in 8 groups of adolescents and young people with Down Syndrome*

	Mean Age	*Logical Thought Test*		*Daily Activities*		*Socialization*	
		Mean score	*Equiv. Age*	*Mean score*	*Equiv. age*	*Mean score*	*Equiv. age*
Group A	24.3	7.3	4.9	285.0	8.5	170.3	7.9
Group B	24.0	11.7	5.7	353.0	16.6	245.0	15.2
Group C	27.7	10.2	5.6	324.5	13.3	228.2	13.0
Group D	23.0	8.2	5.2	280.5	9.6	178.0	7.6
Group E	18.0	9.7	5.5	272.5	8.7	175.5	7.3
Group F	18.0	8.6	5.3	276.0	9.4	205.0	10.6
Group G	19.2	5.5	4.4	222.0	6.1	138.0	4.3
Group H	18.7	6.0	4.5	189.0	4.6	129.5	4.2
OVERALL	21.6	8.4	5.1	275.0	9.6	183.5	8.7

As far as socialization is concerned, we can consider another study conducted in Italy (Moniga, Beschi, & Maeran, 2008), which presents results from a specific rehabilitative plan based on life experience aimed at facilitating "independent life", involving eight groups of children with Down Syndrome. As is shown in Table 2, at the equivalent "logical thinking" age of five, there is superior social adaptability and daily capabilities.

Considering other research carried out in Italy (Ferri, 1989; Ferri, Gherardini, & Scala, 2001; Bargagna, Perelli, Dressler, Pinsuti, Colleoni, Astrea *et al.*, 2004), the results seem to confirm the following tendencies:

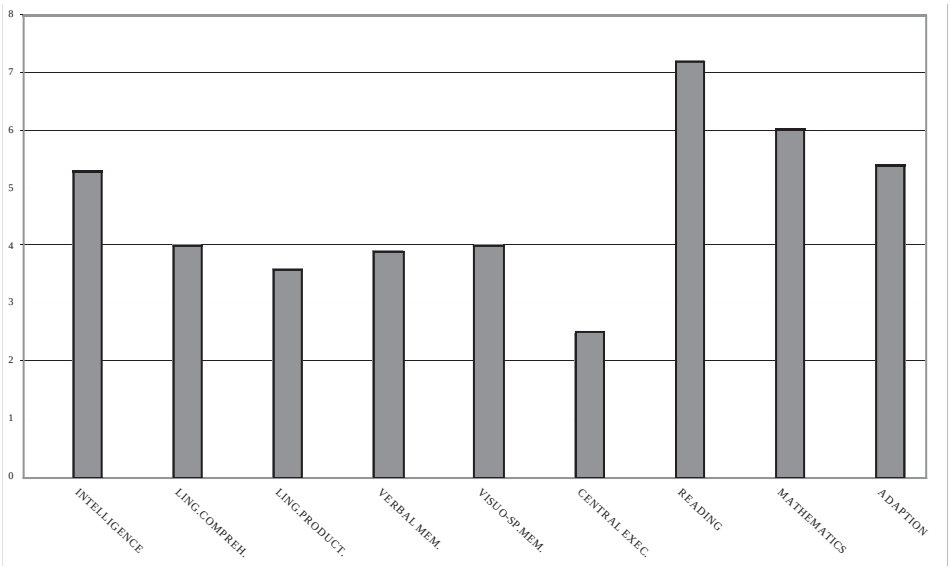
- Up to the age of 11-14, social adaptation tends to reach a similar level to that of children ages 6-7 (and superior to cognitive abilities of one or two years).

- At a higher age, progress has been registered, even if very slowly, up to ages 25-30, which allows for generally average performances for typical developing children of approximately 8 years.

Deficit and surplus compared to Mental Age in Fragile X Syndrome

In a recent unpublished research we conducted with Elisa Moratti, on ten children with X Fragile Syndrome (mean mental age 5 years and 3 months, and mean chronological age, 12 years adn 7 months ranging from 6 years and 10 months to 17 years and 2 months).

Figure 1 - *Cognitive, linguistic, adaptive, and academic performance profiles of 10 boys with Fragile X Syndrome*



As shown in Figure 1 also in this case, surplus relative to mental age is evident in reading performance while less so in maths. Greater still is the surplus (corresponding to adaptive capacity) if the comparison is made with linguistic and mnemonic abilities.

Deficit and Surplus respect to Mental Age in the Cornelia De Lange Syndrome

In a study conducted by Fiori *et al.*, (2008) on eight children with Cornelia De Lange syndrome academic performance parameters tend to be superior to mental age assessed with WISC-R in two individuals from the four taken into consideration (see Table 3).

Table 3 - *Performance of 8 young people with Cornelia de Lange syndrome in the areas of: intelligence, linguistic comprehension and production, visuo-spatial, adaptive ability and academic performance*

Chrono-logic Age	*Mental Age (IQ)*	*Language Understanding*	*Language Production*	*Visuo-spatial Ability*	*Social Ability*	*Academic Performance*
5.5	2.8 (50)	2.8	2.0	2.8	1.6	-
7,6	3.8 (50)	3.0	2.6	3.0	2.0	-
8.9	2.8 (<25)	1.5	1.5	-	1.5	-
10.10	6.4 (64)	4.3	4.3	6.0	4.3	6.0
12.0	11.5 (97)	12.0	9.2	8.0	9.0	12.0
12.3	4.6 (38)	2.9	2.7	5.0	3.2	7.0 (surplus)
15.4	1.9 (<25)	1.5	1.5	-	1.5	-
17.6	13.3 (76)	12.9	11.5	17.6	11.5	14.0 (surplus)

Table 4 - *Performance of 4 adolescents with Cornelia de Lange Syndrome in three different tests (WISC-R, LCO, and CFV) that evaluate different aspects of intellectual performance, and academic performance*

Chron. Age / Mental Age or Equiv. Age	*WISC-R*	*LCO*	*CFV*	*Academic Perfomance*
10.10	6.4	4.11	6.0	6.0
12.0	11.6	6.8	6.6	12.0
12.3	4.6	4.5	5.0	7.0
17.6	13.3	7.0	6.6	14.0

Comparing through different tests the intellectual level of four of the children, in Table 4 we see tests differences in the estimated intellectual level. The test used in this study were WISC-R (Wechsler, 1974), LCO ("*Logical and Conservation Operations*", Vianello & Marin, 1977) and CFV test ("*Correspondence and Functions Assessment*" test, Vianello & Marin, 1998).

These results are particularly interesting if we remember that in the test result the least influenced by environmental factors (educative, social, linguistic) is the LCO test - the CFV and WISC-R tests closely follow. For four persons, academic performance shows great 'surplus' compared to "logical thinking" revealed in the LCO test. Probably this is due to wery high qual-

ity educational intervention, that improved individual skills.. The surplus respect to the CFV test is also important. These results also show a certain tendency to improved performance respect to WISC-R results.

Deficit and Surplus respect to Mental Age in the Prader-Willi Syndrome

Research conducted by D'Amato, Gasparini, Lanfranchi. Moro, Raffa, & Vianello (in Vianello, 2008) offers data pertaining to Prader-Willi Syndrome (table 5). An interesting result is the intra-syndromic variability, comprising individuals with normal intelligence (n. 1), Borderline Intellectual Functioning (nn. 2, 4, 8, 9, 10) and moderate to profound mental retardation. In adaptive behavior, assessed with Vineland Adaptive Behaviour scale, there is a conspicuous surplus in communication and daily skills. Also, in academic performance, both surplus and deficit situations exist.

Table 5 - *Chronological Age, IQ, Mental Age (assessed with LO test), adaptive competence (Vineland test), and academic performance (reading, writing, maths) in 12 children with Prader-Willi Syndrome. The symbols + and – indicate deficit or 'surplus' respect to mental age.*

	Chron. Age	*IQ*	*Mental Age*	*Communication*	*Daily Abilities*	*Socialization*	*Motor Abilities*	*Reading and Writing*	*Maths*
1	5.8	93	5.0	6.1 +	5.0	2.8 -	5.1	-	-
2	6.2	79	4.9	5.11 +	5.1	4.2 -	5.1	-	-
3	6.6	-	2.0-2.6	2.4	2.10	2.3	2.2	-	-
4	8.0	78	5.9	7.7 +	5.0	5.5	4.3 -	-	-
5	8.1	-	1.6-2.0	1.6	2.3	1.6	3.7 +	-	-
6	11.2	47	5.9	6.10 +	9.4 ++	6.4	5.1	-	-
7	11.2	36	4.5	3.10	3.11	4.1	4.1	-	-
8	15.3	70	10.0	10.10	11.10 +	12.4 +	-	4th grade, Elem.	3rd grade Elem.
9	17.0	76	11.11	12.1	16.9 +	15.10 +	-	3rd grade. Middle +	3rd grade Middle +
10	17.8	84	13.6	13.5	15.10 +	11.3 -	-	3rd grade, Middle	3rd grade Middle
11	19.3	54	8.1	9.4 +	10.3 +	14.1 +	-	2n grade Elem. -	2nd grade Elem. -
12	20.0	51	9.8	12.1 +	16.11 +	16.7 +	-	1st grade Middle	1st grade Middle

Discussion

An adequate interpretation of the results presented in this article must assume the existence of a crucial fact: our research did not include any "training" nor selection of participants. This fact is important because the data must be interpreted in the context of a "standard" reality, not of particularly trained persons with mental retardation. Surely, the existence of a "surplus" respect to mental age tends to be present in the studies aimed to evaluate the effects of training or direct intervention (Rynders, 1981; Byrne, Buckley, MacDonald, & Bird, 1995; Rynders, 1999)

But is this also valid, in an international context, for the general population of persons who suffer mental retardation for genetic causes?

We haven't been able to gather convincing nor sufficiently organically-related results for definitive confirmation.

Comparing earlier research results (e.g. Baroff, 1989) reveal that, above all for reading-writing abilities, this "surplus" characterizes the Italian reality more than others, and the crucial variable may be the inclusion of almost all pupils with mental retardation in mainstreaming and not in "special" classes (much different than in many other countries).

At the conclusion of this article, it seems we have formulated more questions than generated answers. The affirmation that 'surplus' in academic and social performance seems to be greater where academic inclusion of disabled persons is more widespread, is still a hypothesis to confirm, however it may be interesting and stimulating.

We also hope that this paper can contribute to promote a comparison between diverse cultural and social realities so that we can better understand how a academic context fostering integration could influence disabled student's performance.

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