1. INTRODUCTION

There exist several hundreds of genetic syndromes leading to mental retardation (MR). Shprintzen (1997) lists more than two hundred genetic conditions conducive to language, speech, and communication disorders. His list includes a large number of MR syndromes of genetic origin. MR of genetic origin represents 30% of all cases of moderate and severe retardation and 15% of all cases of mild mental retardation.

Down syndrome (DS) is the most frequent nonherited condition of MR with an incidence circa one case per 1,000 live births for both sexes (it may be closer to 1/1,500 or less in a number of developed countries due to the conjunction of early diagnostic procedures and abortive practices). No major language difference has been demonstrated between the three main etiologic sub-categories of DS [i.e., standard trisomy 21 (97% of the cases), translocations (2%), and mosaicism (1%)]; except for a statistical superiority in lexical development in mosaic persons keeping up with their tendency to dispose of higher intellectual quotients (IQ).
2. THE SPECIFICITY QUESTION

Given the knowledge currently available and rapidly increasing on a number of genetic syndromes of MR (cf. Hodapp et al., 2000; Rondal et al., 2004), a relevant question is that of the relative specificity or commonality of the major neurobehavioral features of these syndromes. It used to be thought not long ago that the key dimension in MR was IQ — completed by an estimate of adaptive potential. This view, while not being irrelevant, is too general. The scientific approach to MR needs to take the etiologic dimension fully into account. For theoretical and clinical reasons, we need to deepen our knowledge of the various MR entities, starting with the genetic ones, and establish on a firmer empirical basis which neurobehavioral features differ from one entity to another, and to what extent, and which features are found within several or all the MR syndromes to a comparable extent.

3. THE LANGUAGE SPECIFICITY QUESTION

The specificity question must be raised for all the neurobehavioral (as well as the health susceptibility) aspects of the various MR entities. In what follows, I concentrate on speech and language, referring to the major systemic components as usually distinguished in linguistics (i.e., phonology, lexicon, semantics and morphosyntax, pragmatics, and discourse).

Generally speaking, the language of the persons with DS does not contain pathognomonic features (i.e., features that would be found in this condition only). Even outside of the language domain, pathognomonic features in MR syndromes are extremely rare (one
case in point is compulsive overeating found practically only — as a
generic feature — in Prader-Willi syndrome; a genetic syndrome of
MR linked to chromosome 15). Therefore if there is specificity, it will
necessarily be « relative specificity » (some authors speak of
« partial specificity »), i.e., the coincidence in DS of a number of
speech and language features that as a whole, hence systemically,
characterize the entity but do not apply as such to other MR entities.

4. SYSTEMIC SPECIFICITY IN DS LANGUAGE

The speech and language of the persons with DS can be best
defined — in generic terms — as presenting marked formal
deficiencies in conjunction with better preserved semantic and
pragmatic dispositions. I shall not discuss here the noticeable and
at times marked interindividual differences in the language of
The existence of such a variation raises interesting theoretical
questions and should be taken into account clinically, but does not
contradict the general pattern discussed here.

Articulatory and co-articulatory difficulties are the rule in DS,
particularly with the more delicate phonemes in the various
languages. Concurrently, there is most often a slow and incomplete
maturation of phonemic discrimination. This renders the mastery of
the forms of words all the more retarded and difficult.
Morphosyntactic limitations reduce sometimes drastically the length
and formal complexity of the utterances. Inflectional morphology
(tense, concord, number, and gender, pronouns) is often problematic
and variable. Production and understanding of subordinate clauses
and compound sentences are limited. Discourse lacks in cohesion
for the insufficient and sometimes inappropriate use of cohesive tools (i.e., controlled introduction and maintaining of referents, proper use of ellipsis and connecting devices within paragraphs — adverbs, conjunctions, etc.) in order to emphasize the relationships between larger units of discourse.

In contrast, no particular difficulties is observed with the concrete meaning aspects of current lexemes in the language as well as with the basic semantic structures involved in combinatorial utterances (e.g., notions of instrument, agent, action, time and space coordinates, simple transitivity, etc.). These meaning aspects and semantic notions are produced and understood at mental age levels. Additionally, there is socially and contextually appropriate use of the major conventional speech acts (e.g., declaring, requesting in action and information) at mental age levels but with limited formal means. The spatial, temporal, and social deixis and presuppositions are effective at mental age levels. DS persons have access to coherent narrative, descriptive, and argumentative discourses with increase in mental age but again using limited formal means.

5. LANGUAGE ACROSS MR GENETIC SYNDROMES

Illustrative of the perspective on systemic specificity in genetic syndromes of MR is the comparison between DS and two other syndromes nowadays well documented, Williams syndrome—WS—(chromosome 7) and Fragile X syndrome (FXS). The speech and language profile in WS is opposite to the DS one, i.e., better phonological, lexical, morphosyntactical, and formal discursive abilities together with marked difficulties in pragmatics (particularly
with the social and reciprocal aspects of the language exchange).

In FXS, regarding particularly the affected males, the typical profile is intermediate between DS and WS, with phonetico-phonological difficulties (diverse from those typical in DS and displaying dysrythmia, impulsiveness and perseverative speech, disrupted prosody, on the top of phoneme substitution), morphosyntactic and formal discursive (cohesive) limitations, better preserved lexical development, and major pragmatic limitations (e.g., poor topic maintenance and turn-taking difficulties).

A limited number of other syndromes have begun to be studied languagewise (cf. Rondal, 2001; Rondal & Comblain, in press). Some appear to be extremely detrimental to language development and functioning. Such is the case for Rett syndrome (classic) — a neurodegenerative condition related to a mutated gene on chromosome X), Angelman syndrome (chromosome 15), Cat-cry syndrome (Chromosome 5), and Prader-Willi syndrome. Other genetic syndromes of MR are more favorable to language, such as Turner syndrome (chromosome X).

6. CLINICAL IMPLICATIONS

The existence of a language systemic specificity in DS (as well, most likely, as in a number of other genetic syndromes of MR) makes it all the more necessary to reconceptualize our current practices when it comes to language intervention. For example, the remediation priorities in DS as compared to WS are diverse. Typically, DS children need in the first place a language training concentrating on the formal aspects of speech and language whereas children with WS will benefit mostly from remediation
procedures meant to enhance language pragmatic abilities. The same type of reasoning must be applied — in pace with the growing number of specific information available in the published works — to other MR syndromes.

7. THEORETICAL IMPLICATIONS

One reasonable possibility is that the intersyndromic variation corresponds to differences in neurological development and the resulting brain structures. In this perspective, the language systemic specificity in DS must be viewed in close relationship with a number of peculiarities existing in the brain of these persons and the language devoted neurological structures therein.

Examination of DS brains reveals reduction in the weight of the hemispheres, brain stem, and cerebellum; delays in myelinisation, reduction in number of neurons, particularly in some cortical layers. DS persons have reduced synaptic density and abnormal synaptic morphology and contacts, originating in pre- and postnatal stages of neuronal development (Wisniewsky & Kida, 1994).

Studies point to important neurological differences between MR genetic syndromes that may explain the specific fractionation of language functions observed in the phenotypes. Research already suggests that functional differences between DS, WS, and FXS, correspond to syndromic variation at the brain level (Atkinson et al., 1997; Bellugi et al., 1990; Galaburda & Bellugi, 2000; Galaburda et al., 1994); Hagerman, 1996; Jernigan et al., 1993); Reiss et al., 1995, 2000; Wang et al., 1992). It is likely that hypofrontality of neocortex in DS persons together with a reduction in the frontal projections from the corpus callosum is related in significant ways to their
typical poorer verbal fluency and difficulties with the formal aspects of the language functions. In contrast, persons with WS exhibit better preserved frontal, superior temporal gyrus volumes, temporal limbic and neocerebellar structures, together with a selective impairment of the dorsal portions of the hemispheres. This neuroanatomical picture in WS is consistent with the observation of better preserved formal language aspects, marked difficulties with pragmatic language functions (and, beyond language, perceptual—cognitive problems relating particularly to the integration of visual stimuli).

In many male persons with FXS, in contrast again, the decreased size of posterior cerebellar vermis is consistent with the motor deficits, as well as with inattention, hyperactivity, and hypersensitivity to stimuli (Mostofsky et al., 1998), whose speech and language effects are typically observed. Conversely, in FXS, there is enlargement of some brain structures (e.g., the hippocampus) due to higher than normal synaptic densities (probably linked to a defect in synaptic pruning early in brain development; Rakic, Bourgeois, & Goldman-Rakic, 1994). These findings are associated with perseveration and other problems in the executive functions also affecting speech and language regulations in FXS (Abrams & Reiss, 1995).

8. CONCLUSIONS

The preceding analyses encourage the belief that considerable insight into the MR genetic syndromes, and into some of the mechanisms responsible for language development and its difficulties and defects, can be gained from a research perspective
oriented towards empirically specifying the neurological and behavioral particulars as well as the commonalities of the various MR syndromes.

As documented, persons with DS typically present enough distinctive speech and language features to justifying a specific approach in terms of the major objectives for intervention. It is likely, as seen, that these particulars are in close relation with (and are largely caused by) distinctive brain features originating in their particular genetic blueprint. The more we know about these features, the better able we will be to motivate more efficient treatment for the benefit of these persons.

References list


