

# **Current perspectives on genetic dysphasias.\***

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## **Abstract**

The paper documents the major difficulties observed in the oral language development of mentally retarded individuals whose mental retardation is of genetic origin (so-called genetic dysphasias). The extent of inter- and within- syndrome variability is evaluated. Particularly, a comparative analysis of typical language phenotypes in several genetics syndromes is attempted and the possible brain underpinnings of the observed differences are envisaged. Recent cases of favorable language development in mentally retarded individuals are summarized and explanatory variables are discussed.

## **1. Down syndrome**

Mental retardation (MR) of genetic origin represents approximately 30 % of all cases of moderate and severe retardation and 15 % of all cases of mild mental retardation (Aguado & Narbona, 1997). Down syndrome (DS; Lejeune, Gauthier, & Turpin, 1959) is the most frequent noninherited condition (Clarke, Clarke, & Berg, 1985) with an incidence estimated by Dolk, De Wals, Gillerot, Lechat, Aymé, Beckers, et al. (1990) of 1/750 live birth in both sexes. It may be closer to 1/1500 in a number of developed countries due to the conjunction of early diagnostic procedures and abortive practices.

Table 1 summarizes the major data on the language difficulties in DS (for a full review, see Rondal & Edwards, 1997).

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Insert Table 1 here

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No major language difference has been demonstrated between the main three etiological categories of DS (i.e., standard trisomy 21 accounting for 97% of the cases; translocations, 2% of the cases; and mosaicism, 1%; in these latter cases, the embryo develops with a mosaic of normal and trisomic cells), except for a possible lexical referential superiority of mosaic subjects keeping up with their tendency to exhibit higher IQs (Fishler & Koch, 1991).

### **1.1. Prelinguistic development**

Prelinguistic development shows significant delays in DS babies. As a rule, they are less responsive to mothers' verbal stimulations than nonretarded (NR) infants of similar chronological age (CA). Turn-taking skills basic for future conversational exchanges are slow to develop. The type of prelinguistic phrasing that can be observed in NR babies beginning around 3 months of age (i.e., intermittent babbling, approximately 3 seconds long, with phrase-ending syllables lasting longer than other syllables.) is different in DS babies. They take longer to finish a prelinguistic phrase (an average of more than 5 seconds). This extended time frame may explain why mothers and their babies are often found to vocalize simultaneously (Jones, 1977).

### **1.2. The sound of babbling**

The sounds of babbling are mostly similar in types and tokens in NR and DS infants (Smith & Oller, 1981). However, the onset of reduplicated babbling (production of speech-like syllables : “bababa”, “dadada”, etc., is delayed in DS infants) is observed around 6

months in NR infants, versus 8 months in DS infants (Lynch & Eilers, 1991). Reduplicated babbling is a precursor to meaningful speech (Mundy, Kasari, Sigman, & Ruskin, 1995).

### **1.3. Meaningful speech**

Many DS children do not demonstrate consistent use of conventional words before two or three years of age. The frequent shortcomings of in motor development (Rast & Harris, 1985; Wishart, 1988) are likely factors contributing to this delay. Semantic development is also retarded in DS in proportion with the cognitive impairment characteristic of the condition. Some of the early contributors to cognitive development are eye-contact and joint attention on the mother and child's part (Bruner, 1975). DS infants exhibit delays (one month on average) in the onset of sustained eye-contact with the mother, and further delays (two months on average) in the setting of high levels of this behavior (Gunn, Berry, & Andrews, 1982). Delays in imitative, verbal, and gestural abilities (Gutman & Rondal, 1979; Mahoney, Glover, & Finger, 1981; Rondal, 1980; Rondal, Lambert, & Sohler, 1981; Sokolov, 1992) may also contribute to the slower pace of development of meaningful speech productions in DS. Early lexical development shows generally shows a good positive linear relation with MA increase (Rondal & Edwards, 1997). However, as noted by Miller (1999), the rate of new words acquisition of DS children does not keep up to the one of NR children and the equations describing both vocabulary learning curves gradually differ more and more as to slope. The gap continues to widen with increasing age. The existence of noticeable individual differences in rate of vocabulary acquisition among DS children must also be acknowledged. Based on a sample of 43 DS children studied at the University of Wisconsin, using parental report (the McArthur Child Development Inventory), Miller (1999) indicates that 65% of the DS subjects scored below their MA-NR peers whereas 35% were learning vocabulary at a rate consistent with 80% of their MA-NR peers.

### **1.4. The first multiword productions**

The first multiword productions that are not unanalyzed formulae are observable around 4 years in DS children. Mean length of utterance (MLU) is widely used as a criterion variable for assessing language development. Up to a certain level of development almost any morphosyntactic acquisition will be directly reflected in the MLU count. MLU development in DS shows a good linear relationship with CA until early adolescence despite the existence of an important delay (Rondal, 1985; Rondal & Comblain, 1996). MLU values of 1 are usually observed around 2 years. Between 2 and 9 or 10 years, MLU goes from values of 1 to 4 approximately. MLU of 5 or 6 units are generally observed from 12 to 14 years. NR children reach MLU levels of 5 units and more around 6 years. In conversational speech between NR adults, MLU values are often close to 12. The slowness and limitation of MLU development in DS correspond to lasting shortcomings in morpho-syntax. Productive use of grammatical words (articles, prepositions, pronouns, conjunctions, auxiliaries) and morphological marking of gender, number, tense, mode and aspect are limited. Most DS subjects are restricted to monopropositional sentences with correct word order. Subordinate clauses are rare. Even at corresponding MLU levels, DS children may not demonstrate exactly the same kind of syntax as NR peers (Rondal, 1978). For example, when matched for MLU with NR children, DS children tend to use fewer complex verb groups and advanced types of indefinite pronouns. Corresponding limitations can be observed in the understanding of grammatical structures (Bartel, Bryen, & Keehn, 1973).

### **1.5. Pragmatics and communication**

There is a growing literature on language pragmatics in MR (Rosenberg & Abbeduto, 1993, for a review). Young DS children (1-4 years of age) use one-word utterances efficiently to request interesting objects located out of reach (Greenwald & Leonard, 1979). Several studies report few differences between MA-matched NR and DS children in the frequency of speech acts, e.g., question-answer, assertion, suggestion, request, command (Owens &

MacDonald, 1982; Coggins, Carpenter, & Owings, 1983). But important limitations exist in the use of linguistic forms that NR people find appropriate for the expression of particular speech acts. For example, DS children have difficulty in using conventional forms for "softening" their requests or rendering them more "polite" by some formal means such as indirect requesting.

However, topic contribution and topic continuation in DS persons have not been studied in detail. Their turn-taking behavior is systematic and rule-governed. DS have keen desires to keep topics going when conversing and to contribute significantly to conversations. But they often lack the language skills and the relevant knowledge to do so.

### **1.6. Comprehension versus production**

A superiority of language comprehension over language production is often claimed in DS individuals. To a certain extent, a similar situation also prevail in NR people. But what is at stake in this context is the possibility of a genuine discrepancy between language comprehension and production abilities of persons with DS. Miller (1999) reports on the developmental progress in language comprehension and production of DS children (between 1 and 5 years-MA). He indicates that 65% of the total individual profiles reflect greater impairments in language production than in language comprehension.

One may question Miller's suggestion as to the existence of a discrepancy between comprehension and production in DS people on several grounds. Regarding lexical functioning, this may not amount to much more than the usual imbalance between productive and receptive vocabulary repertoires in NR people. Concerning morphosyntactic functioning, it is relevant to recall with Caplan (1993) and Faust (1998) that there are at least three routes to sentence meaning : (1) a syntactic route that computes a full syntactic representation for a sentence and uses this representation to assign aspects of sentential meaning; (2) a heuristic route that uses a reduced syntactic structure (e.g., word order) for the same purpose; and (3) a

lexico-pragmatic (lexico-semantic) route that infers aspects of sentence meaning from single word meaning and knowledge of real-world events. According to the above mentioned authors, the specialized processing mechanisms that assign syntactic structure to sentences are (normally) unique to the left cerebral hemisphere; the right one being able to understand sentences by using semantic relations between single words and world knowledge, that is, by the lexico-semantic-pragmatic route. This being so, Miller's theoretical indication based on the use of global comprehension tests such as the Inventory of Communication Development (Hedrick, Prather, & Tobin, 1984), the Miller-Chapman procedure (Miller, Chapman, Brandston, & Reichle, 1980), and the Test for the Auditory Comprehension of Language - Revised (Carrow-Woolfolk, 1985), compared to results of productive assessment mainly involving free-speech analysis (SALT; Miller, & Chapman, 1993; Miller & Chapman, 1981) and yielding such general measures as performatives, requests for attention or objects, and MLU, may reflect more the use by DS individuals of heuristics 2 and/or 3 above than a genuine morphosyntactic treatment of which most of them may be unable as demonstrated by Rondal (1995) in experimentally controlled tasks of comprehension of reversible passives, relative and causative, temporal clauses, excluding the recourse to heuristic 2 and/or 3 above. Typical DS individuals do not readily produce such linguistic structures, except at times in idiosyncratic expressions; but they fail to understand them their beyond clause level.

### **1.7. Is DS language simply delayed or qualitatively different ?**

The question was raised years ago (Yoder & Miller, 1972) whether language development in MR children is a delayed version of normal language development or whether it shows qualitatively different patterns. It is now possible now to give a more precise answer to this question based on the large number of studies conducted. It appears that language development in DS children is not just a slow-motion version of NR language development. From the early stages on, there are noticeable differences with NR children, including

particular limitations for which adequate explanations are not available yet. Additionally, language development in DS is never complete. It plateaus at various times depending on the particular aspects considered. A strict delay-difference framework is not appropriate for describing language development in MR individuals (Rondal, 1984, 1985, 1987). But language development in DS subjects is not exotic either. The sequence of developmental steps appears to be the same in DS and NR children (similar sequence hypothesis corroborated).

### **1.8. Which language levels are reached by DS adults ? Is there progress beyond adolescence and during the adult years ?**

This question is related to the issue of a critical period for first language acquisition raised by Lenneberg (1967). Lenneberg's conclusion was that due to the maturational calendar of the brain, no basic language development could be possible beyond puberty. He claimed to have observations from DS subjects supporting this hypothesis (Lenneberg et al., 1964). We now have more systematic data pertaining to the problem and are in a better position to assess the validity of the critical period hypothesis for first language acquisition.

In NR individuals, crucial indications are available from Genie, a modern-day "wild child" who was kept away from social contact for most of her first 13 years (Curtiss, 1977). When she was discovered, Genie understood only a few words and did not speak. From that time, she developed relatively rapidly in the cognitive area and enriched substantially her referential lexicon and semantics. But the acquisition of grammatical rules and their use in complex utterances never followed. Her word orders are globally appropriate but the utterances lack bound and free grammatical morphemes. Advanced syntactic devices are missing. Other cases exhibiting the same general pattern have been documented. For example, Chelsea, a hearing-impaired adult of normal intelligence who first attempted to acquire spoken language in her 30s, following successful auditory amplification (Curtiss,

1988), progressed regularly in lexical knowledge thereafter. She scored above the 12-th grade level on the Productive Word Association Subtest of the Clinical Evaluation of Language Functions (Semmel & Wiig, 1980). In contrast, her ability to combine words in utterances remained extremely limited resulting in multiword combinations being ungrammatical most of the time.

Further data suggest a critical period for the development of grammar setting mechanisms. Mayberry, Fisher, Hatfield (1983) showed that individuals who acquired American Sign Language (ASL) as adolescents perform worse on tasks assessing grammatical knowledge than individuals having acquired ASL earlier in life. Corresponding indications have been published by Newport (1990, 1992).

The available data argue in favor of the existence of neuropsychological mechanisms devoted to grammatical and phonological aspects of language. Such mechanisms are tied to the left-cerebral hemisphere and develop according to strong maturational constraints. There is no indication that the development of semantic lexical, pragmatic, and discursive skills (i.e., the more conceptual, social, and informative aspects of language) are characterized by similar temporal constraints.

The critical period for formal language development is likely to have modular characteristics. Expressed differently, there may be an aggregate of particular phenomena coinciding partially in time. The critical phonological period may terminate around 7-8 years of age. Judging from available data on the difference in vocal capacity between congenital deaf individuals or individuals having become deaf early in life (i.e., before about 2 years of age, Menyuk, 1977), and subjects with deafness that occurred later in life, there is probably an earlier critical period for voice setting and control. The ending of the critical period for morphosyntax may be around 14 years. Hurford (1991) suggests that the main determinant



for the end point(s) of the critical period(s) is the "consequences of the interplay of genetic factors influencing life-history characteristics in relation to language acquisition".

Assuming that the evolutionary constraints advocated by Hurford (1991) apply equally to MR individuals (including DS individuals), it can be predicted that no basic formal language development will be readily possible beyond 14-15 years in these subjects. Training effects may still be induced later because critical periods do not end abruptly. But the effect of training will be at an increased cost with advancing CA. Development in the conceptual aspects of language may continue during adolescence and beyond in proportion to a possible continued growth in mental age (MA) (Fisher & Zeaman, 1970, Berry, Groenweg, Gibson, & Brown, 1984).

As reviewed by Rondal and Comblain (1996), the above predictions are supported by facts. Observing language evolution in DS from late childhood onto adulthood, little structural progress is obvious in the phonological and morphosyntactic aspects of language. MLU, for instance, plateaus beyond 14 or 15 years. In contrast to phonology and morpho-syntax, continued improvement beyond childhood is generally the case for referential lexical abilities, pragmatics, and to a lesser extent discursive abilities.

### **1.9. Working memory**

The reference model here so far is Baddeley's (e.g., Baddeley, 1986, 1990, 1992, 1996). According to this model, working memory (WM) is composed of a controlling attentional system (called the central executive) which supervises the activities of two subsystems : (1) the phonological loop devoted to the maintaining of verbal material (auditory-verbal working memory; AV-WM), and (2) the visuo-spatial sketch pad for visuo-spatial material (visuo-spatial working memory; VS-WM). The former subsystem has two components : (1) a phonological store conserving the verbal input approximately 2 seconds,

and (2) an articulatory rehearsal system, based on inner speech, allowing to recycle the verbal material back into the phonological loop, therefore avoiding trace decay.

It is known that WM is impaired in MR individuals (Bilovsky & Share, 1965; Marcell & Armstrong, 1982; Comblain, 1994, 1996, in press). The subjects do not exhibit the modality effect observed in NR children (i.e., auditory presented material is better recalled than visual one) Mackenzie and Hulme (1987) were the first to study the functioning of WM functioning in DS individuals. They reported a reduced WM span in DS compared to NR subjects over a five-years period. In NR children, AV-WM span increases from a mode of 4 items around 5 years CA to 5 items around 7 years, and 5.5 items around 10 years. Several phenomena account for the growth of AV-WM span in NR people : (1) subvocal rehearsal strategy; (2) better organization of the information to be recalled; (3) slower trace decay; (4) faster items identification rate; and (5) faster rate of articulation. Hulme and Mackenzie (1992) investigated articulation rate and subvocal rehearsal in a sample of DS adolescents. They noted a lack of word length effect (NR people usually recall more short words than longer ones within the same interval of time) and an absence of link between articulation rate and WM. These two observations may imply that DS individuals do not use the subvocal rehearsal strategy. Broadley, MacDonald, and Buckley (1995) and Comblain (1996a,b) confirm the absence of rehearsal strategy in DS individual. The three above mentioned studies observed a phonological similarity effect in DS subjects (i.e., phonologically dissimilar words are better recalled than phonologically similar ones) similar to NR people but less marked and not increasing with CA as it is the case for NR subjects.

No matter how limited, the AV-WM of DS individuals can be improved through specific training procedures. Broadley, MacDonald, and Buckley (1994) taught DS individuals to use rehearsal and organizational strategies in order to improve their AV-WM spans. They reported long-term effects of the training. Comblain (1994, 1996a) trained the

use of the rehearsal strategy in three samples of DS individuals (children, adolescents, and adults). She noted a significant increase of the AV-WM span in the trained groups compared to the controls groups. In the trained groups, AV-WM span reached the level expected regarding the mental age. Eighteen months after the training ended, the DS subjects' AV-WM span were still larger than at the beginning of the study.

AV-WM performance may be linked to language development and particularly to lexical acquisition (Baddeley, Papagno, & Vallar, 1988 for neuropsychological data; Service, 1992; Service & Kohonen, 1995; Cheung, 1996 regarding bilingual people; Gathercole & Baddeley, 1990a, 1990b, 1993 for data concerning NR and language impaired children; and Comblain, 1996a; submitted, regarding Ds individuals). It could therefore be relevant in order to facilitate lexical development to increase their AV-WM. Jarrold and Baddeley (1997) claim that the impaired AV-WM of DS individuals can have "important implications for the development of language skills in this population, and in particular, might explain why language development is particularly delayed in comparison to nonverbal abilities" (p. 102).

## **2. Variability across syndromes**

Recent work suggest that language development and functioning may vary consirating across MR syndromes at corresponding psychometrics levels.

Shprintzen (1997) lists several hundred genetic syndromes leading to MR and communication disorders. Relatively few of these syndromes have received detailed phenotypic work and fewer have been studied from a language point of view.

Several language studies have been conducted on Williams syndrome (WS; Beuren, 1972). WS is a congenital metabolic disorder (incidence : 1 case in 10,000 or 20,000 live births) associated with hemizygous deletion including the elastin locus at chromosome 7q11.23. Hemizyosity of the elastin gene accounts for the vascular and connective tissue

abnormalities observed in WS. However, the genes contributing to other features of the syndrome, such as infantile hypercalcemia, dysmorphic facies, and cognitive defects (between mild and moderate mental retardation) remain to be identified (Galaburda, Wang, Bellugi, & Rosen, 1994).

Many WS have good referential lexical abilities (Bellugi, Bihrlé, Jernigan, Trauner, & Doherty, 1990). They seem to have lexical access difficulties in experimental tasks and be less sensitive to word frequencies than normal controls (Tyler, Karmiloff-Smith, Voice, Steven, Grant, Udwin, Davies, & Howlin, 1997). Speech is fluent with correct articulation and prosody. Sentence comprehension and use of morphosyntactic devices are not intact as preliminary indications had suggested (e.g., Pinker, 1994). Recent works (Karmiloff-Smith, Grant, Berthoud, Davies, Howlin, & Udwin, 1997; Karmiloff-Smith, Tyler, Voice, Sims, Udwin, Howlin, & Davies, 1998) show that WS individuals have difficulties in production as well as in comprehension of certain features such as subcategory constraints (e.g., intransitive verbs cannot take a direct object), embedded sentences, and grammatical gender assignment across sentence elements; although their general grammatical skills appear rather good (particularly in adolescents and adults). Discursive ability seems to be relatively preserved in WS subjects (Wang & Bellugi, 1993).

Pragmatics is their area of major weakness. They often have difficulties with topic introduction, topic maintenance, turn taking, and in maintaining appropriate eye contact. WS subjects sometimes appear to be talking nonsense. Their speech may be socially inappropriate and repetitive with incessant questions, and, at times, echoing the interlocutor's sentences apparently with limited understanding (Crisco, Dobbs, & Mulhern, 1988).

Vicari, Carlesimo, Brizzolara and Pezzini (1996) indicated that AV-WM in WS individuals compares well with that of NR subjects at corresponding cognitive levels. Tests

reveal that phonological similarity and word length effects to the same extent in WS and NR subjects. However, WS subjects exhibit a reduced frequency effect.

Gosh, Ståding, and Pakau (1994); Udwin and Yule (1990) suggest that WS individuals have excellent AV-WM and that they tend to store words by mere mimicry. Grant, Karmiloff-Smith, Berthoud, and Christophe (1996) do not agree with this hypothesis. They point out that WS individuals' good language reflects their ability to construct language-specific phonological representation of their native language rather than their good AV-WM.

The syndrome terms "Fragile-X" (FXS; Lubs, 1969), an X-linked disorder passed on through generations, has motivated research studies in recent years (Mulley, Kerr, Stevenson, & Lubs, 1992). The cytogenetic expression of fragile site is on the X chromosome at Xq27.3. It is caused by a null mutation at the FMR-1 gene in which the level of protein in mRNA (messenger ribonucleic acid) is greatly reduced. At the DNA (deoxyribonucleic acid) level, it is characterized by abnormal repetitions of a trinucleotide sequence. FXS is one genetic abnormality following an unusual pattern that is not yet completely understood (Phelps, 1998). Twenty percent of males with the errant gene present no pathological symptom (nonpenetrant). The rest of the affected males are moderately to severely MR (Maes, Fryns, Walleghe, & Van den Berghe, 1994). Approximately, one third of the females are affected with a phenotypic variant of the syndrome determining learning difficulties. A minority is impaired with mild to moderate MR. They are those females having inherited FX from a carrier mother. Surveys of MR population (Webb, Bunday, Thake, & Todd, 1986) suggest that FXS accounts for 2 to 7 % of MR among males. FXS prevalence in the general population is close to .72 per 1,000 males.

Although the situation of the affected and the carrier females is less clear (Dykens, Hodapp, & Leckman, 1994), the language picture for the affected males may be summarized as follows :

1. Speech is fast, repetitive, perseverative, with cluttering and fluctuating rates, increased loudness, and sometimes oral apraxia (Newell, Sandborn, & Hagerman, 1983).
2. Unusual voice effects, dysrhythmia, echolalia, speech impulsiveness, disrupted prosody, and poor intelligibility have been noted (Newell, Sanborn, Hagerman, 1983; Borgraef, Fryns, Dielkens, Pyck, & Van der Berghe, 1987; Gérard, Guillotte, Servel, & Barbeaux, 1997).
3. FXS males frequently omit or substitute vocalic or consonant phonemes (Vilkman, Niemi, & Ikonen, 1988). Utterance formulation is usually defectuous. Receptive vocabulary seems to be relatively preserved (Gérard et al., 1997). Productive morpho-syntax is deficient (Sudalther, Scarborough, & Cohen, 1991).
4. The language of FXS males is pragmatically limited with poor topic maintenance and turn-taking difficulties. They may also exhibit deviant repetitive language (Madison, George, & Moeschler, 1986).

Indications regarding memory limitation in FXS have become available. Dyckens, Hodapp and Leckerman (1987) report a poor performance of all FXS males in memory tasks, particularly in those involving verbal encoding. Freund and Reiss (1991) observe deficits in sentence repetition related to sequential processing limitations.

A few other genetic syndromes have witnessed beginning language study allowing firsts characterization of the major language problems existing therein.

The Cri-du-chat (cat cry) syndrome (CDCS; Lejeune et al., 1963; German, Lejeune, McIntyre, & de Gronchy, 1964) is a rare syndrome (approximately 1 case in 50,000 newborns; Nieburhr, 1978) caused by a loss of chromosomal material from the distal portion of 5p. Twenty percent of the cases are familial with parental translocation accounting for the majority of these. The size of the 5p deletion can vary from the entire short arm to only 5p15. Gersh Goodart, Paszter, Harris, Weiss, and Overhauser (1995) have determined that two

distinct chromosomal regions are associated with differential phenotypic manifestations. Deletions in 5p15.3 result primarily in the high-pitched cry characteristic of the syndrome. The typical facial dysmorphias are lacking and cognitive impairment is mild to moderate. The loss of a small region within 5p12.2, designated the Cri-du-chat critical region (Overhauser, Huang, Gersch, Wilson, McMahon, Bengtsson, Rojas, Meyer, & Wasmuth, 1994) results in the full spectrum of CDCS. Incidence of the syndrome does not vary according to sex (Pueschel & Thuline, 1991). A significant number of individuals reach puberty and survive into adulthood (Niebuhr, 1978). The more typical phenotype presents a characteristic (monochromate) cry at birth, certain dysmorphic craniofacial features with microcephaly, psychomotor retardation, hypotonia, slowed rate of growth, respiratory and ear infections, and frequent orthopedic malalignment. Neurological examination and magnetic resonance imaging (MRI) reveal an hypoplasia of the cerebellar vermis associated with dysgenesis of the corpus callosum. Standard references sources list severe to profound mental retardation, lack of ambulation, lack of speech, and a reduced life as almost inevitable for these individuals. Longitudinal data gathered by Wilkins, Brown, and Wolf (1981) Wilkins, Brown, Nance, and Wolf, (1982) and Carlin (1988a,b, 1990) indicate, however, that the prognosis for most parameters of health, development and longevity in CDCS are much more optimistic today than those presented in common sources of medical information. For example, the risks of major organ anomalies and decreased survival are low. Home-rearing and early intervention are keys to the improved outlook. Data gathered by Wilkins et al. (1982) on 86 home-reared individuals with 5p indicate Mas of approximately 2 years 5 months at CAs around 6 years, with mental quotients inversely proportional to the age at which early intervention began. Carlin (1988b) report on 31 individuals with CDCS seen longitudinally, some of them for 10 years or more, and cross-sectional data from 31 other cases. Cytogenetically, the sample included a majority of terminal deletions, and some interstitial

deletions, mosaicisms and translocations. Carlin notes that in spite of large variations in the size and the location of the chromosome deletions, remarkable phenotypic consistency exists. Early growth failure, microcephaly, significant psychomotor retardation and respiratory and ear infections, are observed in all individuals. Almost 100% of them have hypotonia, at least in the early months. In later years, about 50% retain some degree of hypotonia and experience limitations in their range of motion at certain joints. Attention deficits are general. Most individuals are friendly and enjoy interacting with other people. All CDCS subjects demonstrate cognitive, language and behavioral deficits, but these deficits have been little studied so far. Lack of speech and severe language problems have been noted. However, limited speech and language development occurs in a large proportion of CDS individuals, providing that they are correctly stimulated.

Angelman syndrome (AS; Angelman, 1965) is characterized by neurologic (ventricular enlargement, anomalous cortical growth, abnormal EEGs, and seizure), motor (ataxia), and cognitive. The population incidence is approximately 1 in 16,000 live births (Wiedemann, Kunzen, & Grosse, 1997). Individuals with AS are often found to have severe to profound MR. In 60-70% of the cases, AS is believed to be caused by the absence of maternal contribution (microdeletions and so-called imprinting) to the q11-13 region of chromosome 15 (Christian, Robinson, Huang, Mutirangrera, Liuc, Nakao, Surti, Chakravati, & Ledbetter, 1995). In a small percentage of the cases, the condition is the result of paternal unisomy (inheritance of two copies of the above locus from the father and none from the mother) (Nelen, Van der Burgt, Nillesen, Vis, & Smeets, 1994). In the rest of the cases, no chromosomal abnormalities or unisomy can be detected (Webb, Malcon, Pembry, & Clayton-Smith, 1993).

Quite noticeable in AS is the absence of speech and oral language together with oral motor dyspraxia (Penner, Johnston, Faircloth, Irish, & Williams, 1993). Nonverbal techniques



of communication have been tried with AS individuals but apparently with limited success, suggesting that the capacity for language (not only speech) is impaired in this syndrome (Jolleff & Ryan, 1993; Summers, Allison, Lynch, & Sandler, 1995).

Neurofibromatosis 1 (NF1 - Cole & Myers, 1978; there is a type 2, involving postlingual deafness but no MR, that does not interest us) is a single gene disorder appearing in childhood. The incidence is approximately 1 in every 4,000 live births (Stumpof, Alkesne, & Amieger, 1988). Mutations of the NF1 gene (on chromosome 11) result in abnormal control of cell growth and differentiation of tissues, especially in the central and peripheral nervous systems. Physical features include macrocephally, gliomas and optic nerves enlargement, skin tumors, dysmorphic features, and various neurological problems interfering with functions. Learning disability and mild MR are frequent but there is a considerable interindividual variability (Edridge, Denkla, & Bien, 1989; Dilts et al., 1996). The language skills of children with NF1 are less well developed than those of unaffected siblings with some subjects demonstrating both expressive language deficits and others deficits solely in expressive language, particularly at the level of pragmatic and discourse organization. (Balestri, Lucignani, Fois, & Magliani, 1994; Dilts et al., 1996; Moore, Ater, Needle, Slopis, & Copland, 1994). Fluency disorders including stammering and voice problems are present in 30 to 40% of cases. Lexical reduction and morphosyntactic difficulties may be found too (Saint-Arroman & Slozz, 1998).

Very few studies focused on memory skills in individuals with NF1. It seems that no deficit exists in AV-WM. Zoeler, Rembeck, and Baeckman (1997) mention the existence of visual and tactual memory impairment in adults with NF1. Bawden, Dooley, Buckley, and Camfield (1996) point to nonverbal cognitive deficits, face recalling and drawing complex geometric figures from memory in children and adolescents with NF1.

Klinefelter syndrome (KS; Klinefelter, Reifenstein, & Albright, 1942), a genetic disorder found exclusively in males with an extra X chromosome or two (XXY, XXXY), affects approximately 1 in 1,000 live births (Mandoki, Summer, Hoffman, & Riconda, 1991). KS is characterized by a tall stature, average intelligence or mild MR. Language problems are common, particularly on the expressive side. Auditory processing deficits are characteristic, although language receptive skills are usually within the normal range (Walzer, Bashir, & Silber, 1991). Delayed speech development during early childhood is frequent with prosodic difficulties. Word selection and sentence organization is problematic in some cases (Leonard, Landy, Ruddle, & Lubs, 1974).

Turner syndrome (TS; Turner, 1938) may be particular language-wise. It occurs in as many as 1 per 2,500 live female births. About 60% of the female individuals born with TS are missing one X chromosome, whereas the remainder have a partial X chromosome or a mosaic chromosomal pattern (Ginther & Fullwood, 1938). TS subjects present a reduced stature and abnormal upper to lower body ratio, various affections of the renal and cardiovascular systems, and occasional strabismus. MR is infrequent but specific cognitive deficits appear frequently nonverbal functioning, particularly in visuospatial processing, visual-motor integration and visual memory (Temple & Carney, 1995). Oral language skills are usually preserved and even be relatively strong (Alexander & Monet, 1965; Ginther & Fullwood, 1998). However, Murphy, Allen, Haxby, and Largay (1994) have signalled lower scores in tests of oral language in TS adults with the mosaic TS subjects exhibiting better verbal abilities. Several studies indicate difficulties in written language (e.g., hand writing and reading) (e.g., Pennington, Bender, Puck, Salbenblatt, & Robinson, 1982; Pennington et al., 1985).

Traditional neuropathological measures (EEG, etc.) have failed to identify consistent anomalies in TS individuals (Elliott, Watkins, Messa, Lippe, & Chugani, 1996). However,

brain imagery studies reveal structural anomalies of the brain of TS individuals, i.e.; decreased volumes of hippocampus, caudate, lenticular, and thalamic nuclei, and parieto-occipital brain matter; parieto-occipital asymmetry with left brain regions having greater volumes than right ones in adults (Murphy, DeCarli, Daly, Haxby, Allen, White, McIntosh, Powel, Horwitz, Rapoport, & Schapiro, 1993). These findings are consistent with a "right hemisphere dysfunction" hypothesis in TS (Silbert, Wolff, & Lilienthal, 1977). Clark, Klonoff, and Hayden (1990) have found lower rates of glucose metabolism in the occipital and parietal lobes in TS subjects compared to controls. PET findings by Elliott, Watkins, Messa, Lippe, and Chugani (1996) suggest that parietal and occipital lobes hypometabolism may be common among TS girls with some degree of cognitive impairment but is not evidenced by TS girls without such impairment.

Studies of TS children reveal more visual and spatial processing impairment than verbal processing difficulties (Rovet & Netley, 1982). It could be that VS-WM is impaired in this syndrome (Rovet, Szekely, & Hockenberry, 1994; Buchanan, Pavlovic, & Rovet, 1998a,b).

Prader-Willi syndrome (PWS; Greenswag & Alexamder, 1995) is characterized by dysmorphic features, hyperphagia, hypotonia, and MR. The incidence is 1 in 15,000 to 30,000 live births, equally affecting both sexes (Daniel & Gridley, 1998). The etiology of PWS remains partially unclear. About 60% of PWS is associated with visible deletions in the q11-13 region of chromosome 15. This is the same region as in AS, but in PWS this is the chromosome 15 constituted by the father that is at stake (imprinting) (Butler, Meaney, & Palmer, 1986). Twenty percent of PWS are due to microdeletions in the same chromosomal region, associated with translocations, which can only be detected using molecular genetic techniques (Trent, Valpato, Smith, Lindeman, Wang, Warue, & Haan, 1996). The rest of the cases show no karyotypic defect. In these cases, however, both chromosomes 15 originate

from the mother (Nicholls, Knoll, Buther, Karam, & Lalander, 1989). The above findings argue strongly that PWS is caused by the lack of some paternal genes from region 15q11-13.

Psychomotor development is delayed in PWS. Eye difficulties, particularly strabismus, are noted. By school age, if not before, cognitive, language and behavioral deficits become apparent. Cognitive disability ranges from severe to mild (Dykens, Hodapp, Walsh, & Nash, 1992). Multiple articulation errors, voice problems, and fluency problems markedly lowering speech intelligibility are reported. Oral motor functions, pitch level, and resonance are disturbed and hearing problems are not unusual. Receptive and expressive languages are found to be well below CA as well as language pragmatic skills and communicative efficiency (Edmonston, 1982; Kleppe, Katayama, Shirpley, & Fousher, 1990). Cerebral dysfunction, combined with a characteristic anatomy of the mouth and larynx contribute to altered speech in PWS (Akefeldt Akefeldt, Gillberg, 1997). A tentative profile of the language of individuals with PWS is proposed in Table 2.

Rett syndrome (RS; Perry, 1991; Hagberg, 1993) is another particular syndrome from the language point of view. It is a progressive developmental disorder affecting approximately 1 in 10,000 female individuals. The infant with RS develops within expected limits until 9 to 12 months of age (Kerr & Corbitt, 1994). Regression then occurs drastically affecting language, motor, and cognitive acquisitions. By 7 years of age, RS children are severely MR (Witt-Engerstrom, 1987). Etiologically, the X chromosome is suspected because only females are affected (Trevathan, 1989). In many RS children language rarely develops beyond single word. Hand skills peak between 10 and 12 months-CA (Kerr, Montague, Mils, Ther, & Stephenson, 1987). Many RS subjects do not show behaviors interpretable as indicative of elementary intentions to communicate (e.g., gaze shifts and turns) (Von Tetzchner, 1997). It seems, however, that there are different courses of evolution in RS. In most cases, there is a failure to develop language or the loss of all acquired language during

the regression phase (Coleman, Brudbaker, Hunter, & Smith, 1988; Naidu, Murphy, Moser, & Rett, 1986). However, some girls have a preserved ability to use at least some grammatical language, often with pronunciation difficulties (Gillberg, 1997; Zapella, 1997).

Table 2 displays the language profiles of four genetic syndromes.

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Insert Table 2 here

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As illustrated in Table 2, the language profiles of DS, WS, FXS, and PWS individuals differ substantially in ways not previously stated. The syndrome differences have little to do with the psychometric levels of retardation in each syndrome. Additional research is needed to analyse these comparisons in more detail and to extend the search for other specific (or partially specific) profiles of genetic hypothesis.

One reasonable possibility is that the syndrome variability corresponds to differences in neurodevelopment and brain structures consequential to different aspects of pathological genetic mechanisms. DS subjects are known to have central nervous system dysfunctions secondary to abnormal brain development at the pre-, peri-, and postnatal stages of brain maturation. The examination of DS brains shows a reduction in weight of the hemispheres, brain stem, and cerebellum, delay in myelination, primarily in the development of the association cortex, reduction in the number of neurons in the whole cerebral cortex and more particularly in some cortical layers (Ross, Galaburda, & Kemper, 1984; Wisniewski, 1990; Wisniewski et al., 1996). DS persons have reduced synaptic density and abnormal synaptic morphology and contacts originating in pre- and postnatal stages of neuronal development (Wisniewski & Kida, 1994). The abnormal neurogenesis in DS may primarily reflect genetically determined altered brain programming.

Available studies already point to important neurological differences between syndromes that may explain the specific fractionation of language functions observed in the phenotypes, originating in different genetic bases. Work by Bellugi and associates, at the Salk Institute for Biological Studies, suggest that functional differences between WS and DS individuals correspond to syndromic variation at brain level. Bellugi et al. (1990) compared the neurological profiles of WS and DS adolescents matched for CA and IQ. The WS subjects demonstrated generalized hypotonia, tremor, midline balance problems, and oral-motor and motor abnormalities, suggestive of cerebellar dysfunction. DS adolescents showed minimal hypotonia, little evidence of cerebellar signs, and better performance on oromotor functions. Both groups exhibited equal degrees of microcephaly, cerebral hypoplasia, reduced cerebral volume, and decreased myelination; but the overall brain shapes of each group proved distinct. DS brain exhibit important degrees of hyperfrontality whereas WS individuals have decreased posterior width with reduction in size of the forebrain posterior to the rolandic sulcus, i.e., the parietal, posterior, temporal, and occipital cortical regions; and narrowing of the corpus callosum anterior to the splenium. WS individuals show elongated posterior to anterior length compared to normal brains. Hypofrontality of neocortex in DS subjects together with reduction in frontal projections from the corpus callosum is further demonstrated in a magnetic resonance imagery study by Wang, Doherty, Hesselink, and Bellugi, (1992). These authors relate this neuroanatomical indication to a profile of frontal lobe dysfunction in DS corresponding to poor verbal fluency, perseverative tendencies, and greater difficulty on tasks requiring flexible problem-solving strategies. DS subjects, however, have relatively preserved basal ganglia and diencephalic structures. In contrast, WS subjects exhibit better frontal and temporal limbic structures (Jernigan et al., 1993). There is also evidence in WS of dysregulation of the control of neuronal and glial numbers, as illustrated by increased cell packing density at the cytoarchitectonic level (Galaburda et al.,

1994). This may reflect an interference with naturally occurring cell death and the presence of neurotrophic factors (possibly linked to abnormal extracellular calcium level).

The cerebellar volume in DS subjects is approximately 77% of the equivalent in young normal controls, versus 99% in WS subjects. Although cerebellar size is intact and neocerebellum largely preserved in WS (Wang, 1992), some other neurological findings are suggestive of cerebellar dysfunction. The posterior fossa structures of the WS and DS subjects were further examined by Bellugi et al. (1990), leading to the identification in WS of an anomalous pattern, with neocerebellar vermal lobules showing hyperplasia in the context of low-normal paleocerebellar vermal development and significantly reduced forebrain size. Such an aberrant cerebrum/cerebellum volume ratio could serve to distinguish neurologically WS from other syndromes such as DS (Courchesne et al., 1988). Bellugi et al. (1990) speculate (following a suggestion by Leiner, Leiner, & Dow, 1986, on the possible role of human neocerebellar structures in mental and linguistic functions) that the observed hyperplasia of specific vermal lobules in the context of cerebellar maldevelopment may be related to the language profile of WS subjects. Bellugi et al. (1990) further remarked that their WS subjects were behaviorally grossly similar to unilateral right-hemisphere damaged (normal) adults whereas the DS individuals were more like left-hemisphere damaged aphasics, demonstrating language impairment and a marked tendency to a global processing of the information. The curtailment of the dorsal parietal and posterior temporal areas of the brain in WS subjects, together with the thinning portions of the corpus callosum, may be directly relevant to their visuo-spatial deficits (Galaburda et al., 1994), and indirectly perhaps to their dissociation between AV-WM and VS-WM systems. WS subjects indeed have better preserved AV- than VS-STM, whereas the converse is true for DS subjects (Jarrold and Baddeley, 1997). Similarly, the better preserved size of the frontal and most of the temporal lobes in WS is consistent with the relative preservation of formal linguistic capacities in WS.

Neurological differences in FXS have been little studied so far. They call for cerebral ventricular enlargement and decreased size of the posterior cerebellar vermis in many FXS individuals compared to NR persons (Miller, 1996; Hagerman, 1996). The latter indication is consistent with the motor deficits in FXS (Friefeld & MacGregor, 1993). Decreased amounts of FMRP (the FMR-1 protein) impair the development of the cerebellum Pukinje cells, the cholinergic neurons innervating the limbic system (involved in emotional and mood regulation), and other neuronal tissues (gray matter particularly) that normally exhibit high concentrations of FMRP.

### **3. Within syndrome variability**

Individual language abilities in MR may be distributed according to a Gaussian curve. For each language component, one may expect that a majority of MR persons will score in the central part of the distribution whereas fewer ones will score at the two extremes of the same distribution, the latter cases being either exceptionally favored or exceptionally restricted as to their final developmental level. Table 3 displays summary information on a number of cases of exceptional language in MR documented in the literature (for a full review, see Rondal, 1975, and Rondal & Edwards, 1997).

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Insert Table 3 here

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Additionally, we supply here complementary information on the case of Françoise, a French-speaking DS woman, with normal or quasi-normal formal language abilities (for a full analysis, see Rondal, 1995), as well as several indications from a study still in progress concerning another French-speaking DS woman, named Claudine.



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Insert Tables 4 and 5 here

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The conclusion to the above studies is straightforward. Formal language problems are not inherent in MR/DS qua MR/DS. Rondal (1998) has suggested that the problems of MR/DS persons in the semantic aspects of language, on the one hand, and in the phonological and morphosyntactic aspects, on the other, do not have the same roots. The semantic problems of MR/DS subjects originate in their cognitive difficulties. These problems are unavoidable given the major cognitive limitations defining MR. However, the difficulties of typical MR/DS subjects with the formal aspects of language do not originate in general condition, as demonstrated a contrario by the exceptional cases. They result from particular impairments in the language organization. This runs contrary to the view that all language difficulties in MR are a direct consequence of their cognitive deficit.

How are we to explain the exceptional cases of language development and functioning in MR individuals ?

Particular educational factors (unusual language training procedures by parents, teachers, etc.) do not seem to have influenced the reported outcomes. There is no indication in the cases studied that particular remedial procedures could be held responsible for the advanced abilities documented. Parent-child verbal interactions with MR children have been proven to be basically normal (Rondal, 1978, 1985), when by "normal" it is meant the type and quantity of linguistic input and feedback received by NR children at corresponding language levels. If adaptations of that sort were the key factor in determining exceptional language abilities in MR subjects, one should observed many more such cases.

What about cerebral hemispheric specialization for the language functions ? Dichotic-listening studies have reported a left ear/right hemisphere advantage for speech sound reception in DS individuals (not found in control groups of NR subjects and MR subjects of other etiologies). DS subjects, however, exhibit the expected right ear / left hemisphere superiority in speech production. Elliott, Weeks, and Elliott (1987) have suggested that the language problems of DS persons may be related to a dissociation between the cerebral areas responsible for speech perception and production, causing difficulties of communication between organic systems that normally overlap, and leaving speech reception in control of the right hemisphere that may not be best equipped to handle this function.

The MR language exceptional subjects for whom relevant data are available (i.e., Françoise and Curtiss & Yamada's Laura) are both left-hemisphere dominant for language functions (receptive as expressive). Rondal (1995) has reported corresponding data for 24 DS adults with typical language abilities for DS (15 males and 9 females, aged 21 to 36 years) in dichotic-listening and dual task studies. A large number of these subjects demonstrated interference between verbalization and right-handed movements compatible with the hypothesis of a left-hemisphere dominance for speech production. In the dichotic-listening task, three females exhibited a right ear advantage - from 30 to 70% - (suggesting left-hemisphere dominance). Six males exhibited a right-ear advantage (from 10 to 63%). Retaining those individuals for whom the right-ear advantage was equal to or in excess of 50%, one had two female and one male individuals. These three subjects all demonstrated a positive relative amount of interference in dual task (suggesting left-hemisphere dominance for speech production). They could be considered homogeneous as to cerebral hemispheric dominance for the speech functions. This is also the case for Françoise. However, the language abilities of the above three DS adults were only average for DS persons. Left-hemisphere dominance may be a necessary condition for advanced language development,

[outside of early focal brain lesions determining a transfer of the language control to the right hemisphere at little or no functional cost (Eisele, 1991); a situation that theoretically does not concern MR individuals as they are not supposed to present focal brain lesions as a result of their condition]. It follows that left-hemisphere dominance cannot be a sufficient condition for exceptional language development in MR people.

**Non-linguistic cognitive factors ?** Most MR language-exceptional individuals studied have MAs around 5 years. It could be argued that they are "simply" demonstrating language corresponding to their cognitive level (Moerk, 1994, Bates, 1997). Bates maintains that, in NR children, basic grammatical development is complete by 4-5 years of age (or even before). In NR children. From such a standpoint, she claims, one should expect MR individuals with MAs of 4 or 5 years to exhibit well developed formal language abilities. However, if general cognition at 4-5 years-MA were a sufficient condition for explaining advanced formal language abilities, typical MR subjects with such MAs (and there are many) should similarly exhibit well-developed morphosyntactic skills. But they do not. Typical MR subjects' grammatical development remains largely even incomplete despite at times systematic language intervention.

Alternatively, if one does not accept the idea that grammatical development is complete by 4-5 years of age but insists instead that it goes on until 9-10 years for some structures, then the levels reached by the exceptional MR individuals become impossible to explain in relying solely on general cognitive variables. Indeed, these individuals exhibit grammatical levels much beyond what could be considered to be normal around 4-5 years-MA in this alternative hypothesis. The general cognition hypothesis is contradicted by the data either from the typical MR subjects or from the language-exceptional MR individuals, depending on where one wants to set the time completion of grammatical development in NR children. In our opinion, the language-exceptional MR individuals have more to rely on than

a cognitive level of 4-5 years-MA. They have at their disposal a language specific ability (grammatical) that has been spared in spite of their pathology. This ability is also available to young NR children (around 20-24 months) when they start developing grammar. It is largely lacking in typical MR individuals. All typical moderately and severely MR individuals reach and go beyond 2 years-MA, but as said, they fall short of developing full grammar. Early cognitive development may supply the necessary basis for grammatical development, but it is not sufficient. Needed too are specific devices responsible for the grammatical operations.

The grammatical ability referred to above does not have to be innate in the representational sense out of any logical or biological necessity. It implies, however, the existence of innate architectural constraints (to employ Elman et al.'s terminology, 1996), i.e., a largely innate processing organization dealing with linguistic, and particularly grammatical and phonological representations. Karmiloff-Smith, Klima, Bellugi, Grant, and Baron-Cohen (1995) suggest that there are domain specific predispositions for analyzing language stimuli which with language experience, become increasingly specialized and interconnected. As normal development proceeds, a modular-like organization takes place (weaker than the Fodorian - Fodor, 1983). The former types of modules can be said to be more made than born (Bates, Bretherton, & Snyder, 1988). But judging from the vantage point of the MR literature, fully efficient language modularization, to continue with Karmiloff-Smith's terminology, does not occur in typical MR subjects despite some cognitive, lexical, and pragmatic acquisitions. This again suggests that something else is needed to bring about the modularization process that may be characteristic of advanced language functioning.

Early cognitive functioning is probably needed for triggering or, at least, set the stage for morphosyntactic development. Supportive evidence is found in exceptional MR cases. Françoise, as well as Christopher, O'Connor and Hermelin's subject, Laura, Curtiss and Yamada's subject, and FF, Vallar and Papagno's subject - i.e., those language exceptional

individuals for whom developmental histories are available - were markedly delayed in language onset. Françoise produced only one word (/to/ for couteau, i.e., in French) at 4 years-CA; even worse than many typical DS children at the same age. She developed her formal language abilities between approximately 5 and 10 years. WS children are severely delayed in early language development. It is only when they have a vocabulary size and general cognitive level comparable to those of NR 2-year olds that their grammar "gets off the ground" (Singer et al., unpublished). The above indications suggest that a cognitive-semantic basis amounting to what exists in moderately and severely MR child around 5 years-CA and in NR children around 20 months is needed for the grammatical component to start working when such a component is indeed functional.

What about WM in relation to exceptional language development in MR individuals ? Françoise, Claudine, as well Vallar and Papagno's FF, demonstrated an AV-WM span of 4 digits and more. Their span certainly were lower at the time of their language development. Vallar and Papagno (1993) proposed that FF's AV-WM explains her better formal language abilities. Such a suggestion is not convincing. However, a positive contribution of AV-STM, due to a better functioning of the phonological loop, cannot be ruled out in MR language-exceptional subjects. Françoise, Claudine, as well as FF, exhibit normal-like WM processes when recalling verbal material. They rely on rehearsal strategies based on semi-private speech. Their speech rate is normal or close to normal in contrast to that of typical DS subjects (Rondal, 1995). However, it may be argued that such a contribution of AV-WM in the language-exceptional cases is limited. As said, Françoise's AV-WM span is 4. Her sentence span is 14 words. She is able to repeat correctly sentences containing up to 20 words. This is normal functioning according to data reported by Butterworth, Campbell, and Howard (1986). In repetition tasks, Françoise made few word order errors on sentences containing more than 14 words. Most of her errors were omissions and (trivial) words

substitutions. So were also the typical errors of Butterworth et al.'s university students when requested to recall sentences 15-20 words long. Françoise's immediate recall performance is in sharp contrast with that of typical DS subjects (Rondal, 1995). The latter individuals cannot repeat correctly sentences containing more than 7 or 8 words at best. They frequently omit major sentence constituents. Additionally, and concerning sentence, this time, Françoise has no difficulties in correctly interpreting (center-) embedded subject and object relatives when the relative pronouns and their co-referring nouns are separated by several words. Neither does she experience particular problems when requested to establish pronominal co-reference across sentences in paragraphs with pronouns and co-referring nouns separated by up to eight words. It is reasonable, to conclude that the contribution of Françoise's immediate phonological memory to sentence production and comprehension is a limited one only.

**Variation at brain level ?** Rondal (1998) has suggested that the major determinant of the morphosyntactic and phonological differences observed between typical and exceptional MR subjects operates at brain level. The macroscopic brain structures devoted to the formal aspects of language are probably spared to a large extent in those MR individuals with exceptional language abilities. These structures are damaged and only poorly operational in regular MR subjects. Correctly organized brain macro-structures owe much to the interplay of what Elman et al. (1996) label "chronotopic constraints". This includes constraints on the number of cell divisions taking place in the neurogenesis, relative differences in timing between brain subsystems, differences in synaptic growth according to brain areas and functions. Rondal's (1998) suggestion is that language-exceptional and typical MR subjects markedly differ as to the architectural and chronotopic characteristics of brain development. As indicated, studies of the brain of MR persons reveal major anomalies. The language-exceptional subjects escape the above fate for reasons that may be related to the phenotypic effects of genetics variation. Geneticists agree that there is substantial variation at the genetic

level between people within genotypic categories such as DS, WS, FXS, PWS, and other genetic causes of mental retardation. Most genetic influences on phenotypes are not discrete but polygenic. As a consequence, complex phenotypic traits show a quantitative variation. Many disorders are the result of multiple genes located on one or more chromosome(s). The interaction of these genes determine the expression and the extent of the abnormality (Phelps, 1998). Other sources of genetic variation include variable penetrance of the gene(s), imprinting effects, and the many possible mutations (alleles) that major genes may have (Smith, Pennington, & DeFries, 1996).

Genetic research is yielding more precise gene identification and phenotypic mapping of chromosome 21. Korenberg et al. (1994) have suggested that DS is contiguous gene syndrome. This augurs against a single DS chromosomal region responsible for the DS phenotypic features. DS and its phenotypes are thought as the result of the overexpression and subsequent interactions of a subset of the estimated 1.000 to 1.700 genes located on chromosome 21. Korenberg, Chen, Schipper, Sun, Gonsky, Gerwehr, et al. (1994) have constructed a phenotypic map including 25 features considered typical of DS. They assign a region of 2-20 megabases between regions on chromosome 21 between regions q11.2 and 22.3, as likely to contain the genes responsible for the DS phenotypes. This conception of the genotype-phenotype relationship in DS is consistent with central characteristics of T21, such as the rich variety of phenotypes and the variability in both penetrance and expression of the phenotypic features. It is conceivable that an important within-syndrome variability exists at brain level in language areas of DS persons, consequent on genetic variation. One might suggest with all due precautions regarding linkage analysis with complex behavioral traits additionally to current phenotype/genotype analysis of DS, that there exists a small region in chromosome 21 the triplication, deletion, or otherwise modification of it, is related to

structural abnormalities of the brain in the area responsible for the formal (particularly the morphosyntactic) aspects of language.

The brain-genes perspective defined herein has the advantage of proposing one single type of explanation for the variability observed in the language of typical MR people and the extremes of such variability in the language exceptional cases. It can also be applied to behavioral and brain differences across the genetic syndromes conducive to mental retardation.

The preceding analyses support the belief that considerable insight on genetic dysphasias and on some of the mechanisms responsible for language development, is be gained from additional intervention between the language sciences, the brain sciences, and the genetic sciences. Regarding language and genetic sciences, this interaction is overdue and should be encouraged, as advocated by Shprintzen (1997).



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**Table 1.** Major language problems in persons with Down syndrome

Language component	Semiology
1. Sound articulation and auditory discrimination	<ul style="list-style-type: none"> <li>* <i>Articulatory and co-articulatory difficulties, particularly with the more delicate phonemes.</i></li> <li>- Slow and sometimes incomplete maturation of phonemic discrimination.</li> </ul>
2. Lexical semantics	<ul style="list-style-type: none"> <li>- Reduced lexicon both in number of lexemes and in semantic features within lexemes.</li> <li>* <i>Poor organization of the mental lexicon, both semantically and pregrammatically.</i></li> </ul>
3. Morphosyntax	<ul style="list-style-type: none"> <li>- Reduced length and formal complexity utterances.</li> <li>* <i>Problems with inflexional morphology.</i></li> <li>* <i>Problems with producing and understanding subordinated propositions and compound sentences.</i></li> </ul>
4. Language pragmatics	<ul style="list-style-type: none"> <li>- Slowness of development in advanced pragmatic skills (e.g., topic contribution in conversation, interpersonal requests, monitoring verbal interactions with other people).</li> </ul>
5. Discursive organization	<ul style="list-style-type: none"> <li>* <i>Insufficiently developed discourse macrostructures.</i></li> </ul>

\* The asterisks signal the most serious problems.

**Table 2.** Feature distribution in four mental retardation syndromes

Language aspect	Syndromes			
	Down	Williams	Fragile-X (affected males)	Prader-Willi
Phonetico-phonological	--	++	--	--
Lexical	-	+	+	-
Morpho-syntactic	--	+	-	-
Pragmatic	+	--	--	-
Discursive	--	+	-	?

Key + (+) : relative strength; - (-) : relative weakness; ? : absent or insufficient data

**Table 3.** Exceptional cases of language development and functioning in MR/DS subjects. Studies and Keypoints.

Study	Subjects	CA <sup>1</sup>	IQ <sup>2</sup>	Operational level <sup>3</sup>	MA <sup>4</sup>	MLU <sup>5</sup>	Other language aspects
1. Curtiss et al. (1989); Yamada (1990)	Antony Rick Laura (etiologies unknown)	6 15 16	50  41	Preoperatory Preoperatory Preoperatory	2 years 9 mths		- Correct articulation and phoneme discrimination - Receptive vocabulary at 6 years level - Advance expressive and receptive morphosyntactic abilities, except for Laura exhibiting receptive morpho-syntactic limitations - Semantic, pragmatic, and discursive deficiencies.
2. Seago (1965)	Paul (Down syndrome)	11	60				- Good command of written language expression and reading (average number of words per written sentence varying from 7.14 to 12.50 between 15 and 33 years).
3. Hadenius, Hagberg, Hytnas-Bensch, & Sjorgen (1962); Anderson & Spain (1977); Tew (1979)	Hydrocephalic children	3		Severely impaired cognitive development			- Good ability to articulate, learn words, and use complex syntax - Semantic difficulties
4. Rondal (1994a, 1994b)	Françoise (Down syndrome)	32	60	Late preoperatory to early operatory	7 years 4 mths	12.24	- Correct articulation and phoneme discrimination - Moderately retarded lexical development - Advanced expressive and receptive morphosyntax - Limitations in discourse organization
5. Cromer (1991)	DH (etiology unknown)	Adolescent	35				- Correct articulation - Extensive vocabulary - Complex morphosyntax - Use of standard pragmatic devices in conversation
6. O'Connor & Hermelin (1991); Smith & Tsimpli	Christopher (etiology unknown)	29	67	Preoperatory			- Practice of English within the normal range including complex

(1995)							metalinguistic judgments
							- Good level ability translating in English from 13 languages : French, German, Spanish, Danish, Dutch, Finish, Russian, Greek, Hindi, Norwegian, Polish, Portugese, and Welsh
7. Vallar & Papagno (1993)	FF (Down syndrome)	23	71				- Correct articulation with occasional stuttering like phenomena - Good acquisition of Italian and to a lesser degree English and French vocabularies - Advanced expressive morphosyntax
8. Rondal & Comblain (in progress)	Claudine (Down syndrome)	27	57	Preoperatory	5 years 4 mths	15.39	- Correct articulation - Moderately retarded expressive lexical development - Advanced expressive morphosyntax - Moderate difficulties at discourse level

- Notes.
1. Chronological age in years at beginning of the study.
  2. Intellectual quotient according to standard intellectual scales.
  3. According to Piagetian criteria.
  4. Mental age in years.
  5. Mean length of utterance (computed in number of words plus inflectional morphemes).

N.B. Empty boxes in Table correspond to pieces of information not supplied by the authors in the original source.



**Table 4.** The Françoise (F.)\* case (summary of data)

\* French speaking

1. **CA:** F. was 32 years old at the beginning of the study.
2. **Delay in language development:** F. was producing only one word at 4 years-CA.
3. **Etiology:** Down syndrome (standard trisomy 21: genotype 47,XX, + free 21 in each of the metaphases studied).
- 4.

IQ (WAIS)	Nonverbal	Verbal
Beginning of study	60	71
End of study	64	70

5. **MA:** (Epreuves Differentielles d'Efficiencce Intellectuelle — EDEI):
  - Nonverbal: 5 years and 8 months;
  - Verbal: 9 years and 10 months.
6. **Visual perception** (Test of the "Figures Enchevêtrées" de Poppelreuter): normal.
7. **Left-right discrimination** (Test of Head): performance within normal limits.
8. **Visuospatial and computational abilities:** markedly reduced (e.g., standard scores at the WAIS: cubes: 4; object assembly: 1; image completion: 4).
9. **Visuographic abilities** (Complex Figure of Rey -copying from model-; copying cube and houses in perspectives; Bender-Gestalt Test): difficulties with the macrostructure of the drawings; too much attention to irrelevant details; proceeds by copying and juxtaposing small parts of the model; unable to draw according to perspective; on the Bender-Gestalt test, F. scored at the median note for 6-year-old children.
10. **Expressive gesturing** (Immediate imitation of finger and hand-sequential gestures, after Berges and Lezine's Test): F. scored within the 12 year-old range for most gestural sequences.
11. **Attention - concentration** (Barrage subtest of the KLT Test): F.'s scored 22 (out of a possible 90 points); this places her at the lower percentile of the NR adult population.
12. **Operational level** (Piagetian): intermediate between late preoperatory and early operatory.
13. **Episodic memory** (Paired-associate words; Test of the 15 words of Rey; Cued Recall and Selective Reminding Tasks of Buschke; Complex Figures of Rey: drawing from memory (3 minutes after exposure) reduced in comparison with NR adults but satisfactory delayed recall of verbal material (up to 30 minutes) indicating correct trace consolidation; correct but impoverished delayed drawing.
14. **Semantic memory** (Free association; Fluency tasks): F.'s associations are largely idiosyncratic and prevalently of the syntagmatic type; no evidence of prototypical organisation of common semantic categories (e.g., animal, clothes, transportation means, fruits, vegetables, etc.).
15. **Working memory**
  - A. **Auditory-verbal (AV-WM) span:** 4 units (digits, words, and nonwords)
  - B. **Visuo-spatial (VS-WM) span** (Block-tapping Test): 4 units (surpassed by 92 % of NR adults; compatible with NR children's level around 5 years).
  - C. **Visuo-spatial recognition** (Delayed Recognition Span Test): span: 5.20 (average span of NR adults: 12.08).
  - D. **Visual reproduction** (Weschler's Clinical Scale): score 4 (very low portion of the NR adult distribution; NR population mean: 11.42, SD: 2.76).
  - E. **Basic functioning of phonological loop:** demonstrated phonological similarity, word-length, and articulatory suppression effects; spontaneous and active rehearsal (whispering and/or mezza voce).

16. **Sentence span:** 14 words; at times, F. can repeat correctly sentences containing up to 20 words.
17. **L-Max** (Free speech): 50 words.
18. **MLU** (in number of words + inflectional morphemes): 12.24 (SD: 9.65).
19. **Sound perception and discrimination** (ORL, examination): normal (no auditory loss).
20. **Articulatory ability** (Free speech; Logatomes of Borel-Maisonny): normal.
21. **Speech rate** (Free speech measurement): F.'s overt speech rate varies around 200 words per minute (i.e., approximately 3.3 words -12 to 15 phonemes- per second); this is also the speech rate for normal adults; regular DS adults subjects have overt speech rate varying from 37 to 71 words per minutes (i.e., 1 word -4 to 5 phonemes- per second and less).
22. **Suprasegmental phonology** (Free speech): normal.
23. **Lexical ability -production, referential, comprehension, definition-** (Test de Vocabulaire Actif et Passif; Test des Relations Topologiques; Batterie de l'Aphasie de Liège; Lexical subtest of the Epreuves Différentielles d'Effcience Intellectuelle; Boehm's test of Basic Concepts; Vocabulary subtest of WAIS): productive and receptive levels compatible with nonverbal MA; on the WAIS lexical definition task, the score obtained is one standard deviation below the NR adult population mean.
24. **Expressive morphosyntax** (Free speech analyzed with reference to Halliday's Functional Grammar adapted for French): virtually normal, witness the grammatically correct production of various grammatical types of sentences, including the most structurally complex ones, reflexives constructions, and the correct use of the various obligatory inflectional morphemes.
25. **Receptive morphosyntax** (Specific psycholinguistic tasks): virtually normal; e.g., correct comprehension of subject and object relatives, causal subordinates (with the subordinate clause either preceding or following the main clause), temporal subordinates (with the verbal order of events corresponding to the order of the events in reality or not), declarative affirmative active and passive sentences ranging in plausibility and plausible reversibility, correct use of coreferential mechanism in the case of the anaphoric personal pronouns.
26. **Pragmatic organization** (Free speech): virtually normal conversational skills (e.g., turn-taking, topic distribution, topic continuity, conversational feedbacks and repairs); correctly formulated illocutionary speech acts; correct use of polite forms and indirect requests for action, information, and confirmation.
27. **Discursive organization** (Free speech): discursive organisation (either narrative or descriptive) globally correct; occasional problems with textual cohesion; conjunctive forms such as and, then, but, although, thus, etc., tending to be used more as loose connectors than genuine markers of logical and/or informational dependencies between utterances, phrases, or sentences.
28. **Written language expression** (Free written texts; Dictation): limited and deviant in several respects (e.g., punctuation marking, conventional orthography, narrative macrostructures, morphological inflections).
29. **Reading and comprehension of written language** (Logatomes and conventional words of Borel-Maisonny; School texts; Written Language Comprehension Task from the Epreuve pour l'Examen du Langage of Chevrie-Müller): reading ability is well established although F. is very slow (which contrasts with her fully speed-appropriate oral verbal ability); written language comprehension is at third-grade level, but demonstrates lexical and conceptual limitations.
30. **Metalinguistic abilities**
  - A. **Phonological awareness** (10 subtests orally presented: selecting or producing rhymes, isolating initial or final phonemes in target words, fusing phonemes into words, spelling words, etc.): F. is able to segment common French words into syllables (but very slowly and at time with some degree of overlapping between neighboring syllables). She cannot regularly identify separate phonemes in words.

**B. Sentence judgement and repair** (for grammaticality and semantic acceptability): F. is able to detect and correct word order errors appearing in grammatically incorrect but semantically appropriate sentences. She is also able to detect and mend grammatically correct but semantically abnormal sentences. However, she did not detect inflectional morphological mistakes.

**C. Grammatical analysis** (Active declarative affirmative sentences presented in written form): F. can often identify main verbs (actional as well as nonactional) and grammatical subjects, direct objects, indirect objects, time circumstantial elements, and locative circumstantial elements (in asking the school-type questions: qui- who, quoi- what, à qui - to whom, à quoi - to what, quand - when or où - where). She could perform the above analysis on monopositional sentences only and, in some cases, on main clauses pertaining to complex sentences but leaving subordinate clauses unanalysed.

**31. Cerebral hemispheric specialization**

**A. Dichotic listening** (Directed attention procedure): REA (right-ear advantage) or LEA (left-ear advantage) calculated from the following formula computed for each ear:

$$Dichotic-listening\ score = \frac{(30-E1) * 100}{30}$$

Where 30 is the number of syllables presented to each ear and E1 the number of intrusion errors.

F.'s REA = 63%, suggesting LHD (left-hemisphere dominance) for speech reception.

**B. Dual-task study** (Finger-tapping task combined with sound-shadowing): Relative amount of interference averaged per second (RAI index) evaluated with a formula comparing experimental steps.

F.'s RAI = +4.05, indicative of an interference between verbalization and finger-tapping that is more marked for the right hand; suggesting LHD for speech production.

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**Table 5.** Claudine\* - Study in progress

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\* French speaking

1. **CA:** C. was 27 years old at the beginning of the study.
  2. **Etiology:** Down syndrome (standard trisomy 21).
  3. **IQ** (WAIS): nonverbal 57, verbal 61.
  4. **MA** (EDEI): nonverbal: 5 years and 4 months; verbal: six years and 3 months.
  5. **Nonlanguage cognition:** important weaknesses in spatial, numerical, and time cognition; preoperatory level on Piagetian measures.
  6. **Working memory:** AV-WM span: 4; V-VS-WM span: 4.
  7. **Speech rate** (articulation speed): below NR but > to typical DS individuals.
  8. **Oral language production:**
    - Lexicon: EVP 68%; not very different from typical DS adult individuals;
    - MLU (words + inflection morphemes): 15.39;
    - Grammatically correct expression of temporal, causal, and relative subordinates in free speech; her speech on the whole is more parataxic than that of Françoise.
  9. **Written language:**
    - Correct writing and reading abilities, albeit very slowly;
    - Correct marking of subject - main verb or subject - auxiliary concord; correct use of the obligatory inflections on the nouns, verbs, adjectives, and pronouns.
  10. **Metalinguistic ability:** seemingly clear phonological awareness.
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