Friulian Journal of Science **5.** 2004, 85-91

Another way from behavior onto the brain and the genes*

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1. Introduction. Since the last part of the 19th century (the observations of Paul Broca), we know that language functioning depends on particular cerebral structures.

Over something like the latter 25 years, we have come to realise that particular genes are involved in linguistic development. For example, in the most complete review of research on the subject to date (more than one hundred genetic studies of language – twin, adoption, and linkage studies), Stromswold (2001) shows that genetic factors account for much of the variance between people with language disorders and some of the variance in linguistic abilities among normal people. There appear to be genetic factors that specifically influence linguistic abilities, in addition to heritable factors that influence both nonverbal and verbal abilities. Furthermore, some studies suggest that different genetic factors are involved in different aspects of language (e.g., semantic vs. syntactic abilities). Stromswold (2001) concludes: "Taken together, the results of adoption and twin studies support the hypothesis that language is partly the result of innate predispositions and structures specific to language" (p. 705).

Stromswold's review has generally been taken to mean that there exists an empirical basis for the brand of inneism put forward by the Chomskyan school in linguistics and psychology (cf. Pinker, 1994, 1999), i.e., so-called representative inneism which postulates that a number of supposedly universal concepts such as *noun*, *phrase*, *verb*, *grammatical subject* (of), *object* (of), *transformation move-alpha*, etc., are encoded genetically and contribute significantly to the specifically human blueprint for language development. Actually, nothing in the data summarized by Stromwold as well as in other corre-

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* Lectio Magistralis, Third Annual Congress of the Friulian Scientific and Technological Society, Gorizia, October 2004.

sponding pieces in the literature, guarantees that such an interpretation is the correct one. It is as likely (more likely in my opinion) that the genetic effects uncovered in the studies mentioned operate indirectly, i.e., through the mediation of particular devoted brain structures which then are responsible for the behavioral phenotypes. In other words, the cause-effect relationship may not be from linguistic genes directly to some particular linguistic knowledge or lack of it in some disorders, but from particular genes to language devoted brain structures and then to linguistic knowledge, development and behavior (normal or abnormal). In the first case, there is no language acquisition in the strict sense; the role of the brain is "simply" to select a particular grammar (the one matching the linguistic input received) among a number of possible ones normally coded in the genes. In the second theoretical case, there is language construction through the particular operations of some brain structures (innately predisposed to perform so) acting on the linguistic input received during a number of years until the resulting system matches the one of the mature people in the verbal community.

2. Levels of language rapresentation. Clearly, then, one of the most interesting problems in the neurosciences today has to do with the specification of the three-way relationship between: (1) *linguistic functioning* (and linguistic development in children), i.e., language behaviors; (2) *cerebral structures* devoted to the language function (and their maturation in children); and (3) *language genotype*, in the sense of the particular gene set whose effects determine the setting of the cerebral structures allowing language development and functioning; at least until it can be proven that there is a part of the genotype that carries linguistic information, which so far has not been demonstrated and, in my opinion, is very unlikely (see also Elman et al., 1997, and others, for an equally skeptical view on representational inneism in language development).

In order to contribute to the clarification of this three-way relationship, we need to dispose of precise information on each one of the levels involved as well as their particular ways of interacting with each other. A drastic limitation regarding the sciences of languages (functioning) is that it is not permissible to carry provoked experiments in order to test particular (hypotheses regarding the structures involved (for example, reducing or eliminating all or parts of the linguistic input to the baby, removing particular areas of the language brain, favoring genetic mutations or silencing specific genes, etc.). The language pathologies, although humanly extremely regrettable, have helped considerably (from the time of Broca), constituting in some respects "natural" (i.e., unprovoked) experiments. Encouraged by Ribot, Professor at the Collège de France, in the last part of the 19th Century, the so-called pathological method, has been extremely useful and is far from having yielded all its resources.

Since the time of Broca until the 1950ies, it is overall the aphasias that

contributed most to our knowledge of the organic basis of language. From the 1960ies on, the influences of the evolving fields of psycholinguistics and generative grammar have had a strong influence on the acquisition research and the study of the language pathologies in supplying models of development and organization of the language function. The studies on the cerebral basis of bilingualism and multilingualism have brought new perspectives on the ways the language brain organizes itself and is disorganized in pathological states. Noteworthy also in recent years, the studies regarding developmental dysphasias in children (i.e., specific language difficulties in children with normal cognitive abilities). In some cases, retrospective inquiries have encovered the existence of language pathological predispositions running in families. Some common genes have been identified and suspected to be responsible for the problem. Once more the question is whether such pathological genes influence directly development because they carry linguistic information indispensable for normal language development or whether (and more likely, in my view) they negatively affect brain development in some aspects critical for development.

A different research perspective, which has been of great interest to me for the last ten years or so, regards the intersyndromic differences in language development and functioning in persons affected by a moderate or severe form of mental retardation with a genetic origin.

Most people know of Down syndrome (trisomy 21), or "mongolism", as it was labelled for a long time following the erroneous suggestion of Langdon Down (the English physician who was the first to publish a systematic description of the condition in the last part of the 19th Century). The chromosomic etiology (presence of three chromosomes 21 instead of two) was ascertained by the French geneticist Jerome Lejeune and colleagues, in the early sixties. From that time (and already before), numerous studies have been published regarding language development and its difficulties in Down syndrome persons. Following the great progresses witnessed in the field of molecular genetics in the last quarter of the 20th Century, a large number of genetic syndromes leading to a moderate or severe degree of mental retardation have been identified (more than 400; Moser 1992; Shprintzen 1997); only a limited number of which have begun to be scrutinized regarding cognitive and language abilities.

Among them:

- Williams syndrome (abnormal chromosome 7)
- Fragile-X (chromosome X)
- Cri-du-chat (chromosome 5)
- Prader-Willi (chromosome 15)
- Noonan (chromosome 12)
- Angelman (chromosome 15)
- Neuro-fibromatosis type 1 (chromosome 17)
- Turner (chromosome X)
- Klinefelter (chromosome X)
- Rett (chromosome X)
- Rubinstein-Taybi (chromosome 16).

One significant aspect, as discovered in recent years, is that the typical

language profile differs in the syndromes; i.e., if one looks at the typical abilities of the persons affected with the above syndromes regarding major language components (phonology, vocabulary, morphosyntax, pragmatics, and discourse), he finds out that the profiles (relative strengths and weaknesses) are different from one syndrome to the next and this at corresponding cognitive levels. This suggests that purely cognitive abilities are not determining variables regarding intersyndromic language variability.

Let's illustrate the above statement with some schematic indications regarding three syndromes (Table 1).

Table 1. Intersyndromic differences in language functioning in three genetic syndromes of mental retardation (Rondal, 2004).

Language components*	Down syndrome	Williams syndrome	Fragile-X sindrome (male sujets)
Phonetics-phonology	– –	++	– –
Lexicon	–	+	+
Morphosyntax	– –	+	+
Pragmatics	+	– –	– –
Discourse	– –	+	–

* Key: + (+) relative strength; –(–) relative weakeness.

2. Research perspectives

Where do we go from here? It so appears, even if the research in that perspective has only barely begun, that there exists a link between the language difficulties encountered in moderate and severe mental retardation and various genotypic abnormalities. These abnormalities are (in a way) unspecific. I mean that there does not seem to be a direct association between such and such language limitation in the retarded persons (e.g., particular phonological, semantic, syntactic, or pragmatic problem) and discrete pathological genetic characteristics (e.g., particular aberrations at the genic or chromosomic level).

A reasonable hypothesis would be that a whole number of pathological genetic influences can gravely disrupt the neurogenesis but that they do it in different ways which are relevant for explaining particular behavioral deficits. If this is so, we should be able to observe syndromic differences in neuroanatomical (and neurophysiological) aspects that may be linked to particular language profiles. Intersyndromic research in neuroanatomical aspects and neurogenesis of mental retardation has barely begun and there is still a long way to go. The available data, however, suggest that mentally retarded persons with various genetic syndromes do differ in various aspects of brain organization. For example, a trend of studies, initiated by Bellugi and colleagues at the Salk Institute, in San Diego, USA,

with the aim of comparing brain anatomy in Down syndrome and Williams syndrome adolescents matched for chronological age and intellectual quotient, has shown that although both groups exhibit equal degrees of microcephaly, cerebral hypoplasia, reduced cerebral volume, and decreased myelination, the overall brain shapes in each two group are distinct. Down syndrome brains present important degrees of hypofrontality whereas Williams syndrome individuals show relative preservation of anterior cortical areas but have decreased posterior width with reduction of the forebrain posterior to the rolandic sulcus, i.e., reduction of the posterior parietal, temporal (with relative preservation of mesial-temporal, however), and occipital cortical regions. Individual with Williams syndrome have elongated posterior to anterior length compared with normal brains, a greater ratio of frontal to posterior (parietal-occipital) tissue, and disproportionate volume reduction of the brain stem. Hypofrontality of neocortex in Down syndrome subjects, together with reduction in the frontal projections from the corpus callosum, is also demonstrated in studies of magnetic resonance imagery. The observations can be related to a profile of frontal lobe dysfunction in Down syndrome, corresponding to perseverative tendencies, difficulties on tasks requiring flexible problem-solving strategies, poor verbal fluency, and particular difficulties with the formal aspects of language (see Rondal 2004, for more detail).

The cerebellar volume in Down syndrome subjects is approximatively 77 percent of the equivalent in young normal controls, versus 99 percent in Williams syndrome. A particular cerebrum/cerebellum volume ratio (i.e., marked by reduced forebrain size in the context of largely preserved neocerebellum) could serve to distinguish Williams syndrome. The better preservation of brain posterior frontal areas and neocerebellum in Williams syndrome could explain their better verbal abilities particularly regarding the phonological and morphosyntactic regulations (see, for example, Fabbro et al. 2000, regarding the possible role of human neocerebellar structures in linguistic functions).

Neurological differences in other genetic syndrome of mental retardation have been less studied yet. In many males with Fragile-X syndrome, cerebellar abnormalities are observed. Decreased amounts of FMR-protein (the protein linked to the mutated gene characteristic of the condition) impair the development of the cerebellum Purkinje cells and other neuronal tissues (grey matter, particularly) that normally exhibit high concentration of FMRp. These findings may be associated with perseverations, stereotypies, hyperactivity, attentional impairment, and other problems in regulating motor activity reverberating difficulties on the language sphere (particularly regarding the production aspects) (cf. Hagerman 1996, and more recent works). Still other neuroanatomical indications regarding other genetic syn-

drome of mental retardation could be supplied (regarding, for example, Rett, Klinefelter, and Turner syndromes) (Rondal 2004).

4. Conclusions

Where does that leaves us? Please notice that I am not implying that the neuroanatomical (and neurophysiological) peculiarities encovered (and those currently being discovered in this research field) are sufficient explanations for the language characteristics of the persons affected with the syndrome studied. I am not even arguing that they are explanations in the strict sense, for they could be parts of more extensive causal circuits and subsystems within the brain.

What I believe to be of particular interest is the research perspective outlined, i.e., the systematic setting in close relationship of the three determining levels of language functions: behaviors, devoted brain structures, and genetic underpinnings.

I believe it to be most likely that brain structures are not mere depositories of predefined formal linguistic knowledge (even of a very general, perhaps universal, type) but that devoted structures within the brain do actually construct this knowledge analyzing particular language input during the development years. Whenever the brain structures do not develop adequately (in the many ways that they can go astray) – for example, but of course not uniquely, for genetic reasons), normal language acquisition cannot proceed and particular pathologies occur. Advancing along the way indicated above, will only be permitted by collaborative research efforts from behavioral and neuroscientists and geneticists.

The empirical indications already at disposal (and those to come) have (and will have) important implications for the clinical and rehabilitative work in at least two ways: first, the particular profiles of relative strengths and weaknesses in the genetic syndromes of mental retardation (here regarding the language functions) immediately translate into particular intervention programmes and rehabilitative priorities (for example, the major difficulties with the formal aspects of language in the context of better preserved pragmatic dispositions in Down syndrome; the reverse profile in Williams syndrome; the particular need for systematic speech training and control of dysfluencies in the context of favorable lexical development in males with Fragile-X syndrome, etc. Second, a deeper knowledge of the relationship between language behavior and development, on the one hand, and the two regulating levels, brain and genotype, on the other, will allow eventually to specify better the degrees of freedom that one has in any rehabilitative endeavor with a mentally retarded person, from a better knowledge of the neurobehavioral plasticity left over in a given pathogenetic condition.

This in no way will restrict or nullify the importance of behavioural clinical work with these people. But it is of great importance to assist and guide the clinical enterprise with a solid neurogenetic framework.

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