Visuospatial abilities in Prader Willi Syndrome

P. Alen, O. Vega, S. Martínez, P. Copet, D. Thuilleaux, J. Jauregui.

In recent years the study of genetic syndromes has gained in importance with the promise of a 'cognitive genetics', augured by some authors as directly relating genes and conduct. While still far from this new science, the enormous number of studies on the topic has produced a notable advance in the development and understanding of models of brain function. Furthermore, these studies have shed some light upon the complex evolutive processes that mediate the relation between genes and conduct.

The Williams Syndrome has commanded much attention, mainly because of the spectacularity of its first descriptions, which described it as "showing intact linguistic abilities" in the face of a "general mental retardation" with a special weakness for visuo-spatiality. This irregular phenotype suggested a direct gene - conduct relation, especially when it was considered that specific genes were known to be affected.

With its particular phenotype, the Prader Willi Syndrome also appears interesting in this respect, especially in the light of the fact that some individuals demonstrate special abilities which enable them to solve some visuo-spatial tasks, such as jig-saw or word-search puzzles, accurately, even at levels higher than mental-age controls. Below I will attempt to explain these special characteristics from an evolutive point of view and in relation to the hypothesis of central coherence.

Recent genetic studies have described problems in the 15q11q13 zone - commonly deleted in Prader-Willi subjects - in the presence of savant capacities, such as the ability to carry out calendar calculations.

A deeper analysis of the visuospatial abilities in PW shows the presence of some alterations in visuospatial systems, and it is possible that these alterations could lie behind this high function in some particular tasks. An example of this would be the WS, where an abnormal semantic organisation would enable them to name familiar objects faster than same-age controls, but, on the other hand, also produce a longer latency in the naming of objects when semantic specifications increased.

The "central coherence" model of autism spectrum disorders is an interesting one. According to this, autistic individuals would present difficulties in the integration of information in a globally coherent way. The processing style of these individuals would be characterised by a local bias predisposing them to attend to certain details, which, while manifestly a handicap, might aid them when solving tasks requiring such a processing style.

The strategy used by PW to solve jigsaw-puzzles also appears to be very special. Almost without looking at the model, they would tend to begin at the borders and methodically complete the structure in this way, keeping the unarranged pieces near each other. This strategy would minimize working memory requirements, as well as planning and global-processing ones, making possible successful task completion.

Accordingly, a strong correlation between results in a figure copying task (Rey figure) and some measures of visual working-memory has been found in our studies, but no relation between this latter and figure organisation (structuration) measures. This would

explain how this local strategy could help some individuals with sufficient visuospatial working-memory capacities achieve nearly normal levels in a figure coping task, but not enable them integrate the whole figure. Integration would, in my opinion, be prevented by an insufficient working memory fractionating perception in its initial stages. However, a step-by-step accumulation of these small units would produce a nearly normal appearance.

This 'local strategy' argument has been used to support two opposite findings: the relative success of PW and the failure of WS on the same tasks. Subjects with WS, while supposedly using a 'local strategy', find these tasks very difficult. Their copies of the Rey figure are so fragmented that they draw the different parts of the figure as if they were different drawings, set apart on the paper. Measures of their visual working-memory are below those of PW. Thus a filtering role for working-memory could be hypothesized at the entrance of the perceptive system making impossible all integration below a certain level. Above it, however, partial integration might take place, sufficient, in fact, to understand the figure as one arranging small pieces of it together, though insufficient to perceive its global structure with its different parts. Thus, as in the case of PW, small fragments would be set close together like jigsaw-pieces giving a global appearance of what is really a fragmented one.

Similar integration deficits have been found in numerous syndromes associated with developmental problems and it is hypothesized that their cause could be the lack, or alteration, of connexions between different parts of the brain or amongst them. An example would be the visual system. This would produce a fragmented storage of information exemplified by the difficulties in 'top-down' recovery of verbal memories. For instance, these subjects would tend not to profit from clues such as "it was an animal" to the same extent as normal controls when remembering a list of words. Similar observations in WS seem to indicate the same. When asked to describe an object, they would tend to enumerate characteristics such as colour or size, finding it very difficult to say to which class it belongs to (for example, "it is an animal").

PW children are also described by their parents as being especially fond of jigsaw-puzzles and word searches, interests linked to obsessive traces such as their concern for order, symmetry and exactness. These motivational characteristics, in conjunction with a use of a 'local strategy', might contribute to the differential development of some abilities, which would surpass others through development, despite sharing the same starting point.

This theoretical reflection, while still vague and lacking in empirical confirmation, should not be disregarded as a merely theoretical proposal, but used as an instrument to integrate, better understand and recall the everyday functioning of PW individuals. For instance, recognizing their integrating problems could allow us to help these individuals arrange the information they somehow receive. They could, for example, be given small units of information, given the categorical rules, and multimodal (intersensorial) information delivering strategies etc. used.