

DOWN SYNDROME PHENOTYPE

Precise descriptions of Down syndrome

In recent years there has been much research interest in looking for behavioural phenotypes (or specific profiles of strengths and weaknesses) that are associated with specific conditions – particularly conditions with genetic origins such as Down syndrome.

This kind of information may be very helpful in alerting parents and professionals to the particular difficulties a child may have. It may also lead to the development of different approaches to treatment or interventions for children with different conditions – even when they all have

similar global levels of delay as measured by mental ability tests. This is the view of Deborah Fidler and colleagues in a paper in which they review the information on the language and communication development of children with Down syndrome, Williams syndrome or fragile X syndrome. They present a range of evidence supporting the view that there are distinct patterns of abilities and disabilities associated with each of these conditions.

Taking Down syndrome as an example, they then go on to illustrate how this information could be used to guide 'syndrome-specific' intervention strategies. Their suggestions for children with Down syndrome include strengthening oral-motor and speech development from the babbling stage onwards, thinking about ways to improve underlying processes which may be having an impact on speech and language learning such as verbal short-term memory and using strengths in visual memory to support learning, using signing as a bridge to talking and targeting 'means-end thinking' (early problem solving skills). They strengthen their argument with a review of some evidence that children in these specific groups do respond differently to intervention strategies and with a call for more of the research community to take an interest in developing and evaluating interventions.

Continuing on the specific phenotype



Using visual learning strengths to teach reading

theme, an experimental study from Joanne Roberts and her colleagues reports on the receptive vocabulary, expressive vocabulary and speech production skills of boys with fragile X, boys with Down syndrome and typically developing boys matched on non-verbal cognitive abilities. The boys with fragile X were further divided into those with or without autistic profiles.

Boys with Down syndrome showed more delay in receptive vocabulary and speech production than boys with fragile X and no autism but not in expressive language. The boys with Down syndrome showed significant delay in all 3 aspects of communication when compared with typically developing boys, supporting the evidence of specific speech and language impairment for boys with Down syndrome – that is speech and language skills that are poorer than expected in relation to their non-verbal cognitive ability.

The gap between cognitive ability and expressive vocabulary was greatest for the youngest children and disappeared at the older ages. The authors suggest this may be due to social confidence and life experience leading to accelerated vocabulary learning and use over time.

The study also found an effect of maternal education on both expressive and receptive vocabulary – that is mothers with more education tended to have children with larger vocabularies – reminding readers that there are considerable social

influences on language and communication learning. The authors note the need for interventions that focus on increasing speech accuracy and well as word learning for boys with Down syndrome and also stress the need to assess each child as an individual.

One danger of pursuing the 'phenotype' model in research studies is that it leads to a belief that phenotypes, linked to genetic disorders, cannot be changed but are an inevitable outcome of a condition. Both these papers take the positive view that we can use the information on the expected differences in early develop-

mental phenotypes in order to be more effective in our interventions and possibly then reduce the expected weaknesses for the next generation of children with these conditions. There is already some evidence that this is possible^[1,2] but much more is needed.

Sue Buckley

Original research papers

Fidler DJ, Philofsky A, Hepburn SL. Language phenotypes and intervention planning: Bridging research and practice. *Mental Retardation and Developmental Disabilities Research Reviews*. 2007;13:47-57.

Roberts J, Price J, Barnes E, Nelson L, Burchinal M, Hennon EA, Moskowitz L, Edwards A, Malkin C, Anderson K, Misenheimer J, Hooper SR. Receptive vocabulary, expressive vocabulary, and speech production of boys with Fragile X syndrome in comparison to boys with Down syndrome. *American Journal on Mental Retardation*. 2007;112(3):177-193.

1. Buckley S, Bird G, Sacks B. Evidence that we can change the profile from a study of inclusive education. *Down Syndrome Research and Practice*. 2006;9:51-53.
2. Buckley SJ, Bird G, Sacks B, Archer T. A comparison of mainstream and special school education for teenagers with Down syndrome: Implications for parents and teachers. *Down Syndrome Research and Practice*. 2006;9:54-67.