Down Syndrome News and Update 4(4), 135-139
Research

The development of the Down syndrome phenotype

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In the first of these new features, Mandy summarises a discussion between the Trust's practitioners, which focused on a new research paper by Deborah Fidler entitled 'The emerging Down syndrome behavioural phenotype in early childhood'.[1]

This month's practitioners meeting involved an interesting and fruitful discussion of a new paper published in *Infants and Children*. The author, Deborah Fidler, of Colorado State University has been working in the field of genetic syndromes associated with intellectual disabilities, including Down syndrome since 1997.

This paper clearly and concisely reviews the growing body of research into the already well-established concept of the behavioural phenotype associated with Down syndrome.[1,2] However, the major focus of the paper is to examine the profile from a developmental perspective, i.e. whether the so-called 'typical' (and some might say 'inevitable') profile presented by the majority of individuals in childhood and adolescence is already present and/or becoming established in infancy. She frames this question within the context of early intervention, highlighting the necessity of creating a curriculum and style of delivery, which is grounded in scientific research evidence regarding children with the specific diagnosis of Down syndrome and not a programme that simply fits a wider target audience of children with developmental delay.

This review does not aim to describe the typical profile as Sue Buckley reviewed this very clearly in her article 'Specificity in Down syndrome' in the last issue.[4] Instead, the aim is to draw out the conclusions of Deborah's paper with regard to the development across time of the phenotype and some of the implications of this for early intervention and school support.

Key points in Deborah's article

- Research supports an emerging Down syndrome phenotype:
- visual and social functioning and self help and daily living skills are relative strengths
- speech and language, verbal processing and motor functioning are relative weaknesses
- Certain aspects of this phenotype are apparent in infancy although the differences between the skills areas are much smaller than in older children
- The discrepancies between skill areas may be magnified as the children age and develop
- Intervention in infancy may be able to prevent or offset the developmental trajectory associated with Down syndrome
- Intervention which is grounded in scientific research into the phenotype and utilises the children's strengths is likely to be most effective

What is a 'phenotype'?

The term phenotype describes sets of observable traits, as determined by both genetic makeup and environmental influences. Deborah explains that phenotypes are 'probabilistic'.[pp.87] This means that people who have Down syndrome are *more likely* to share one or more characteristic specific behaviours, as opposed to those with a general developmental delay. Secondly, not every child with Down syndrome will show all the behaviours and thirdly some of the behaviours associated with Down syndrome may appear in other syndromes. Phenotypes are purely descriptions of characteristics, which often appear together as a syndrome. The phenotype does

http://www.down-syndrome.info/library/periodicals/dsnu/04/04/

not explain why these traits appear together or how they might interact.

The development of the phenotype

Deborah indicates that from her own research using the *Mullen Scales of Early Learning* that a typical profile of "relative strengths in visual processing and receptive language and relative weaknesses in gross motor skills and expressive language skills" appears to be emerging by the age of 2. Further data from parent reports showed the toddlers to have strengths in socialisation skills but weaknesses in communication and motor skills.[p.94] Interested readers are referred to her original paper to explore the specific details of each domain of functioning as Deborah highlights many pieces of research which show that sweeping generalisations about strengths and weakness are not always useful as there are many subtle nuances within these areas, i.e. when more specific skill areas are examined, one begins to see that not every skill within a domain is always a strength or a weakness. For example, within visuo-spatial processing (generally discussed as 'a strength'), although visual imitation and visual memory skills do seem to be strengths, spatial and visuo-construction skills appear to be relatively weaker.[p.88] There are also aspects of social communication, which are weaker than others, e.g. non-verbal requesting (using gestures like pointing to 'ask' for something) is a relative weakness whereas social interaction and engagement skills are often seen as a strength.[pp.90-91] Again, in the field of motor functioning, running and agility are actually sometimes on a par with typical development whereas other motor skills such as motor planning and certain precise movements of limbs and fingers are more delayed.[pp.91-92]

Through the looking glass: Tiny variations and huge gulfs

Another very important point, which Deborah describes, is that although the typical profile does appear to be present in an embryonic form during infancy, the discrepancies between areas of strength and areas of weakness are much smaller than they appear to be in older children. She suggests that these "tiny variations" may become "magnified" as the children mature and "snowball" into the distinct profile with which many readers will be familiar. The implication is that certain interventions, at so-called *sensitive periods* during development may help to "prevent or off-set" this process.[p.87]

The concept of critical and sensitive periods is a controversial one in the research literature, not least due to the fact that the term 'critical' implies that once a certain window of opportunity has passed, one can never acquire a skill with the same degree of competency. However, it is likely that there may be times when the brain is more receptive to certain types of learning. It is likely that there is, for example, a sensitive period for learning grammar and this is particularly salient for children with Down syndrome, for whom this appears to be a particular area of weakness.[see 4 for more information] This led the team to discuss the potential of early reading for developing children's verbal short-term memory, syntactical knowledge and use of grammar in their expressive language. We discussed the fact that those children who had started reading activities early at the age of 2.5-3.5 years of age and had developed extensive sight vocabularies may have made better progress with regard to the effects on speech and language, particularly grammar development, than those who started later. Interested readers are referred to *Reading and writing development for individuals with Down syndrome - An overview* [6] for a discussion of these findings. This is, of course, not to say that targeted intervention at any age is ineffective, as many adolescents and adults with Down syndrome have demonstrated significant progress in a wide range of skill areas, including literacy, later in life.[6] The old adage 'better late than never' comes to mind even if 'it's never too late' is not always quite the case.

Nature and nurture

An extremely important point raised by this article, which must be highlighted to every individual working or caring for a person with Down syndrome, is that a diagnosis of Down syndrome in early infancy does not predict the future in any certain way. Development is a dynamic process, whereby experiences within the environment are key to the way in which the brain itself develops and is organised.

The idea of a typical profile associated with Down syndrome is important to gain a general understanding of which styles of intervention may be helpful however, each individual with Down syndrome will only share the typical traits to a greater or lesser extent, some may share characteristics of other syndromes, some may have a full diagnosis of another syndrome, as Sue Buckley describes in her article on dual diagnosis, see p. 114 of this issue. As we shall see in the section on implications, below, this point is crucial with regard to the ways in which parents and professionals interact with the children as no single approach will be equally effective for every individual with Down syndrome.

Early intervention

Above it was noted that intervention in infancy may help to prevent or offset the emergence of some of the key weaknesses associated with

Down syndrome in childhood and beyond. However, Deborah indicates that this can only happen under certain circumstances, possibly the most potent of these being the importance of tailoring the intervention to the emergent profile of strengths and weaknesses presented in infancy.[p.94]

Ideally, intervention should start as early as possible and should always actively involve parents and children. Also, it is likely that parents who understand the rationale behind different aspects of the intervention will be more successful in their targeted work at home. Therefore, some degree of overt training may be necessary as well as modeling practical activities and games to the parents. The activities are likely to be most effective when they build on the children's visual and social strengths and are tailored to the individual likes and dislikes of the individual children. One way in which this idea can be applied is by embedding opportunities to display skills which are likely to be more difficult for the child (e.g. speech sound production or precise fine motor movements) within tasks that will be perceived by the child as 'do-able' and fun. For example, action songs or pretend play scenarios, which are often extremely popular, could be used as a context in which to practice particular speech sounds.

Also, it is important to collect information from families about activities, toys, topics and so on which will be particularly motivating to each individual child so that these can be incorporated into activities to target specific skills and as reinforcers (rewards). One can begin to see from this discussion that for early intervention for children with Down syndrome to be most effective, a collaborative approach between practitioners and parents is essential.

Personality and motivation

Deborah also draws the reader's attention to research on personality and motivation, which indicates that many children with Down syndrome share certain qualities, which manifest themselves in a characteristic style of social interaction.[pp.92-93] Although the literature on temperament in infancy suggests that infants with Down syndrome show the same distribution of temperaments as typically developing infants, by childhood there seems to be a tendency to show decreased task persistence and variable motivation during work-related situations. Many children can be seen to utilise their strong social skills to engage others in 'diversions' and collude with them in a variety of behaviours unrelated to the learning task. She also cites research that indicates that children with Down syndrome tend to make more efforts to elicit help from adults than to persist with a task and develop their independence and problem solving skills and a study, which suggests that children with Down syndrome are less likely to repeat previous successful outcomes.[p.93] These observations may be helpful in developing guidelines to help those who support and particularly are involved with testing and assessment of infants and children with Down syndrome.

However, it was noted above that children with Down syndrome show enormous individual variation. As practitioners, an understanding of the behavioural phenotype is extremely useful for those working with infants and children with Down syndrome at one level, (e.g. curriculum planning) but on an individual level, it is also crucial to take a step back from the profile and simply observe the subtlety of the behaviours displayed by the child and the exact situations in which they occur.

Conclusion

This paper provided an interesting starting for point for a discussion, which helped us to reflect on our practice with children and families both in early intervention and school liaison. It also provided ideas for future publishing and training activities and inspired us to think about potential research questions for the future.

We would encourage parents and practitioners to read the full article which has in-depth discussion of issues relevant to early intervention for all aspects of development and can be found at http://depts.washington.edu/isei/review/iyc.html.

The Trust practitioner and research team look forward to sharing Deborah's work with her further at the *4th International Conference on Developmental Issues in Down Syndrome*, to be held in Portsmouth, in the UK, in September, 2005. Please see page 159 for more details of this forthcoming conference. The full programme is also now available on our website, http://www.downsed.org

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ISSN: 1463-6212

Research Research Research

We are keen to support postgraduate research into issues relating to Down syndrome and this section of *Down Syndrome News and Update* often includes adverts from researchers who are currently recruiting participants. Readers should note however, that the Trust are not usually involved in any way with these projects and therefore with have no further information about them.

In a future issue of *Down Syndrome News and Update* we are hoping to publish a short article about the advantages and disadvantages of participating in research projects. This will include information that families should consider when deciding whether or not to participate and make suggestions about questions to ask the researchers before making a decision. It will also give information about the ethical guidelines that have to be upheld by those completing research with children and families.

Language Research Project – help needed

The psychologists at The Down Syndrome Educational Trust and the Child Development Group, Psychology Department, University of Reading are collaborating to look at early language development in children with Down syndrome, children with autism and typically developing children.

We need parents willing to complete a straightforward checklist of the words and signs that their child understands and/or uses. If you have a child with Down syndrome between 2 years and 8 years of age and would be willing to help with this, please contact me on 02392 893885 or at sue.buckley@downsed.org. I will then mail you the questionnaire and stamped return envelope.

The findings will be very helpful in adding to our knowledge of the rates and variability of progress in children with Down syndrome as well as contributing to the Reading research project.

We will send the results to everyone who takes part.

Professor Sue Buckley, Director for Research
The Down Syndrome Educational Trust

the down syndrome
educational trust
http://www.downsed.org/

Early Support

Information for parents – Down Syndrome

The staff of The Down Syndrome Educational Trust and the Down's Syndrome Association have been working together with the Early Support team to produce an information booklet which will be given to every new family with a baby with Down syndrome in the UK. The booklet will be available from mid-July and you can order a copy by going to the website at www.earlysupport.org.uk

We would like to thank the many families who have worked with us and read two drafts as the booklet progressed. It is quite a detailed booklet and we appreciate the time given by everyone who read it and sent back comments. They were all noted and the final text reflects parents' views.

This booklet is part of a whole programme of work funded by the UK government and aimed at improving services to families for the first three years of a baby's life. We are all working on more materials to support early intervention services, including a Developmental Record for Children with Down syndrome for families, which will be described at our September Conference and available spring 2006.

You can read more on the whole Early Support programme, which firmly believes in putting control in the hands of families and in developing flexible and effective support services, on the website. Family Support packs can be obtained from the website and, if you live in the UK, you might ask if key-worker services are being implemented in your area yet. Each family should have a key-worker to help them to decide what they would like for their child and to co-ordinate early intervention and therapy. In some areas, this is being called 'The team around the child' approach. For all our readers from other countries, all the materials are available for download from the website. This is an exciting initiative and is certainly having an impact on services to families in the UK.

Early Support
Helping every child succeed
http://www.earlysupport.org.uk

Research Research Research

Requests for participants

What works for us: Parental perspectives and Early Intervention

Alice Paige-Smith and Jonty Rix

There is currently a great deal of emphasis on children with Down syndrome and their parents becoming involved in Early Intervention programmes. These interventions can involve all kinds of professionals and activities and can be supported by many different kinds of advice. Much of this Early Intervention involves parents carrying out specific tasks with their children, other activities are built into a play situation, some involve professionals and require little parental involvement.

This piece of research wishes to find out what approach parents feel best suits them, their child and their situation. The first stage of the research project has considered the types of provision and early intervention experiences that parents have had with their children. Subsequently, we would like to focus on parents views - how you think your child learns in the home context. It is intended that this should operate as a pilot study for a wider research project into parental perspectives on Early Intervention.

The research is being conducted by Alice Paige-Smith and Jonty Rix from the Open University. Alice has considerable experience of presenting the parental viewpoint, while Jonty is a parent of a young boy with Down syndrome. The research would involve either a 30-60 minute face-to-face interview or a phone interview.

If you are interested, please get in touch by e-mail or phone:

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Jonty Rix 01444 819210
E-mail: j.r.m.rix@open.ac.uk

Exploring learning through listening

Much of schooling involves learning through listening to teachers and it is important that they are able to recognize the diversity of pupils' needs and adapt their speech accordingly. I know from my experience as a teacher that this is not an easy task. My research project therefore aims to provide classroom teachers with a better understanding of what speech rate is most appropriate to use in the classroom to help improve the education of children with Down syndrome ages 5-11.

The project has 3 stages:

1) The initial stage will involve collecting information through observation and assessment to establish the learning profile of each child with Down syndrome. The results will then be used to decide which story is best suited to each child in the later listening tasks.
2) The second stage will involve presenting the stories at slightly different speech rates and seeing what pupils can remember.
3) For the final stage, the results will be analysed to establish the optimum speech rate to help the children. Staff in the pupils' class will then be asked to try and adopt that speech rate during some lessons. The interaction between teachers and children will be observed and this will be followed up by interviews with the class teachers.

In order to make this research worthwhile I need to find a broad sample of children with Down syndrome. I would therefore be most grateful if you would be willing to allow your child to join this project. If you are prepared to allow your child to help with this research, please contact me by 'phone, letter, fax or e-mail.

Thank you very much.

Jin-Chang Huang

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