New and old directions

Frank Buckley

Improved social and educational opportunities and access to informed healthcare are helping today's generations of people with Down syndrome to achieve more and live longer. This progress is bringing new challenges. Scientific research is steadily improving our understanding of the condition. Future improvements in the lives of people with Down syndrome will require multidisciplinary efforts and more applied or translational research with practical outcomes. In this context, this journal has reviewed the roles it plays in communicating research to specialists and non-specialists, families, practitioners and researchers alike.

Life has changed for many people with Down syndrome in many places around the world. Improved social and educational opportunities and access to informed healthcare are helping current generations of people with Down syndrome to achieve more and live longer. The families, clinicians, researchers and associations that have campaigned and worked hard to deliver improved opportunities over the past 30 years deserve much credit for this. And the thousands of people with Down syndrome who (when given the chance) have repeatedly risen to new challenges and shown themselves to be more able than previously thought possible, deserve the utmost respect, admiration and continued support.

Research aimed at improving our understanding of Down syndrome has progressed alongside these advances (and often supported them). Investigations of language development, reading abilities, numeracy, speech, hearing, memory and social development inform modern teaching practice and early intervention for young people with Down syndrome[1-4]. Clinical studies of the health issues more frequently associated with Down syndrome inform modern healthcare guidelines[5,6]. Meanwhile, our understanding of the biology of Down syndrome is improving, helped by advances in the fields of genetics, molecular biology and neuroscience[7].

Modern healthcare guidelines and equal access to treatment are helping life expectancy to approach 60 years in some places. Meanwhile, in many countries, the numbers of children born with Down

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syndrome each year is remaining steady, despite the increased availability of prenatal testing. Combined, these factors are leading to more people alive with Down syndrome[8]. Meeting their needs is an important social and political issue.

As with many aspects of human life, the picture is not the same throughout the world. Despite improvements in some countries, life for many people with Down syndrome in many countries is very difficult and exclusion, deprivation and discrimination are all too common. The recently adopted *UN Convention on The Rights of Persons with Disabilities* (*News*, p.5) is a step in the right direction at governmental level and community efforts in some places are leading to positive change (*News*, p.6), but much work remains to be done.

Nor is progress in generally progressive communities uniform. Educational practice and opportunity often varies between providers purportedly operating within a common legal and educational system. Obstructive and disinterested institutional disregard for the rights and needs of young people with Down syndrome may be on the decline, but it is still too common and remains a cause of lowered educational outcomes for young people with Down syndrome and anxiety and distress for their families. Even for those not tackling institutional discrimination, many challenges remain. The skills, expertise and resources necessary to properly support the education of people with disabilities are often in short supply in today's education systems. Indeed, our understanding of just what constitutes a first-rate educational experience for young people with Down syndrome is by no means satisfactory.

Reappraising our roles

Against this background of widespread (though by no means universal) progress for people with Down syndrome, the emerging challenges of growing and ageing populations, the practical challenges facing those tasked with providing effective developmental and educational support and healthcare, and continued advances in many areas of scientific endeavour relevant to people with Down syndrome, this journal has reappraised how it fulfils the roles it plays in supporting progress.

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First published in 1993, *Down Syndrome Research and Practice* has always sought "to bring research to parents and practitioners... attempting to draw out the practical implications of research"[9]. At first predominantly focused on language and cognitive development, our subject matter has broadened over the past 14 years. At the same time, we have probably not worked hard enough to make this broader range of content more accessible to the non-specialist. Since 1998, this journal has had a sister publication – *Down Syndrome News and Update* – that has focused more on providing overviews of research for a broad readership and presenting practice-focused case studies and reviews. This has separated information about practice from the research content of this journal. We believe that research scientists need to be in touch with practice and the everyday challenges facing people with Down syndrome and their families just as much as practitioners need to be informed of current research evidence to support their work. We are therefore discontinuing the publication of *Down Syndrome News and Update* and reabsorbing the best practice content back into *Down Syndrome Research and Practice*.

New content

To successfully blend original scientific reports (that can be quite technical to the non-specialist) with examples of good practice, news and reviews in ways that meet the needs of non-specialists and specialists from a variety of scientific and non-scientific backgrounds is a challenge. To help achieve this, we have adopted a number of new sections within the journal to present information in different ways. Original reports of scientific research studies will continue to form the foundation of our scientific content. Where these present group studies, they will be grouped as *Reports* and where they involve one or a few individuals they will be presented as *Case Studies*. Accompanying original study reports will be *Reviews*, *Opinions*, *Hypotheses* and *Perspectives* – each contributing in different ways to the scientific literature. Contributions published in each of these categories of original scientific content will continue to be subject to the peer-review process. (Authors should refer to the revised publishing policies and guidelines at www.downsed.org/research-practice/authors).

To report and summarise international news, research advances and progress in practice, we are also introducing *News*, *Research Highlights*, *Updates*, *Practice*, *Essays*, *Resources* and *Guidelines* sections. *Research Highlights* will offer brief reports of notable recent research findings published elsewhere. *Updates* will offer more detailed reports of progress in research and practice, either published in this journal or elsewhere, presented for a non-specialist readership. *Practice* reports present the observations of families, carers and practitioners and may examine any aspect of development, health or education for one or more individual. *Essays* offer personal reflections on topics of interest to our wider audiences, and will include the reflections of people with Down syndrome. The *Resources* section will include reviews of books, materials and other resources of interest to families, practitioners and researchers. *Guidelines* will include recommendations for good practice. These will usually be the product of appropriate community groups, organisations or committees – for example, the Down syndrome medical interest groups.

Not every section will be present in every issue, though every issue will carry news, research highlights, updates, practice and scientific papers. We have colour-coded the sections to assist readers in finding particular content. As well as better defined sections, we are also introducing more extensive use of glossaries and diagrams to improve the accessibility of all of the content of the journal. We are also adopting full colour production throughout.

Online improvements

Print is no longer the primary publication medium for any journal and we are making substantial changes to improve the quality, availability and functionality of our online editions. *Down Syndrome Research and Practice* forms a substantial and important part of the new *Down Syndrome Online* web site. From the publication of this issue onwards, we will publish online simultaneous with or in advance of print publication. We are also improving the indexing and linking of the journal online. We have joined CrossRef (www.crossref.org) and all of our content will be assigned Digital Object Identifiers (DOIs) and registered with CrossRef to assist the cross-referencing of cited papers and the easy location of online resources. We are also accelerating indexing in MEDLINE/PubMed with the electronic submission of bibliographic information immediately on publication.

Open Access

This journal supports improvements in the lives of people with Down syndrome by communicating new scientific knowl-

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edge and evidence-based practice to the widest readership. To maximise our reach, we are adopting Open Access publishing policies. In short, these will ensure that all of our content is freely accessible online and can be freely reproduced for non-commercial purposes (for more detail and precise legal terms, please refer to www.down-syndrome.org/open-access).

We will continue to produce and offer subscriptions to printed editions. We hope that those who value the added convenience and benefits of printed copies will continue to subscribe. We also hope that those who utilise and value the online editions will occasionally pledge a donation to support ongoing publication. The Open Access publishing model offers alternative revenue streams to subscriptions, but we still expect that this journal will continue to require substantial charitable subsidy.

Multidisciplinary coverage

Over the past 20 years, scientific and technical advances have opened whole new areas of research of relevance to people with Down syndrome. Down syndrome begins with the presence of an additional chromosome 21 (or in a few cases, a part of it) at the time of conception. From this point forward, what we see as Down syndrome is the result of the complex interactions of genes and environment. Understanding the mechanisms set in motion by the additional copies of chromosome 21 will illuminate how some things work differently in people with Down syndrome. Improved understanding of the genetics, molecular biology and neuroscience involved in Down syndrome will inform the characterisation of the particular issues associated with the condition. This knowledge will contribute to the further improvement of developmental and educational interventions. Over time, it may lead to useful pharmacological or gene therapies – indeed, a recent study in mice offers hints of possibilities in one of these areas (*Updates*, p.20).

Unravelling the complexity of human development requires explanation at many levels (from the cellular mechanisms of genes to the psychological characteristics of behavioural traits). Inevitably, these explanations will become more entwined. The importance of increasingly interdisciplinary approaches to behavioural research is often noted.

This journal's overriding aim remains to keep a diverse readership informed of current progress in our understanding of all aspects of Down syndrome. We will increasingly seek to highlight and present research from across the full spectrum of scientific research relevant to our understanding of Down syndrome at all levels.

From the laboratory to the classroom and the clinic

As fascinating and exciting as contemporary behavioural science is, proof of advances for people with Down syndrome is found in classrooms, clinics and everyday life. It is here that the work of researchers, practitioners and families comes together to make a difference.

There are many aspects of teaching practice, speech and language therapy, clinical care and family life that are fundamental to the developmental progress, health and quality of life of people with Down syndrome. Yet, practical evaluations of interventions under 'real life' conditions are scarce. Whilst such study designs may not be ideal for examining some theoretical issues, they can be of immense practical importance. Determining what does and does not work in today's classrooms, with real teaching staff and current resources is critical if we are to advance teaching practice.

This journal will welcome, encourage and highlight such studies. By presenting accounts of practice from practitioners and families, we also hope to interest researchers in pursuing applied or translational research with practical outcomes.

Developmental complexity

Many aspects of human behaviour are substantially heritable (that is, many differences are substantially due to genetic factors) and there is no reason to suppose that this is not the case for people with Down syndrome. No doubt some of the wide variation in outcomes for people with Down syndrome is explained by individual genotypes (including individual third copies of chromosome 21).

However, this does not mean that environmental differences are of little consequence for human development. Indeed the recent history of people with Down syndrome offers a striking example of the consequences of deprived environments and the effects of reducing that deprivation. It is clear that differing educational and developmental support, peer groups and social circumstances influence outcomes for people with Down syndrome (as for all of us).

We are yet to determine what people with Down syndrome are able to achieve, given the most effective support. The achievements of many young people with

Down syndrome today would have been exceptional relative to those seen 40 years ago. Whilst the gains from escaping institutions have now been realised for many, the benefits to be achieved from properly resourced and supportive inclusive education, and from accepting and supportive communities, are still incomplete. How exceptional today's achievements will seem in another 40 years is hard to tell (*Research Highlights*, p.9). There clearly are limits (as there are for all of us) and on many measures it seems likely that these limits will be in the lower range of typical human achievement, but it is still not clear precisely what they will be.

This journal will remain focused on improving our understanding of development and the evaluation of developmental and educational interventions. To this extent, we are reaffirming the directions originally set out in our very first issue –

> *"We think that we live in exciting times and that the future for many children and adults with Down syndrome is brighter than ever before. Research is uncovering many of the reasons for their delayed development and practitioners are using these insights to develop effective teaching and remedial strategies. At the same time social attitudes are changing and our children are being accepted and included in mainstream society. These changes will bring progress but not without throwing up new problems to solve along the way. We hope that this journal will act as a forum to inform on progress and to discuss issues which need to be solved."*[9]

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