

# What do we know about the needs of children with Mosaic Down syndrome and their families?

Sue Buckley and Gillian Bird

**Mosaic Down syndrome is rare and families feel lost when they receive this diagnosis – often weeks or months after the child's birth. A new association will lead to progress.**

In October 2003, the first international conference on Mosaic Down Syndrome will be held in Florida, USA, following the recent establishment of an International Mosaic Down Syndrome Association. The establishment of the Association is an important step, for both parents and professionals, as we do not know enough about the effects of Mosaic Down syndrome. The IMDSA website (see below) has provided parents with a chance to share experiences with other parents and if many families worldwide join the Association it will be possible to begin to gain more detailed information on the needs of these children and their families. At present, it is difficult to find enough children in any locality to study their development. The reader is referred to the personal case histories of children on this site as they make clear the uncertainty that families feel when they receive the diagnosis and the very varied developmental profiles of children with Mosaic Down syndrome.

## A rare condition

It is estimated that 2 or 3 children in every 100 children diagnosed as having Down syndrome have the Mosaic form. Children with standard Down syndrome have an extra copy of chromosome 21 in every cell and this is the usual profile for some 96 in 100 children with the diagnosis. Children with Mosaic

Down syndrome have some cell lines with the extra chromosome and some cell lines that are not affected and have the usual 46 chromosomes. A few children, 1 or 2 in a 100, have the translocation form of Down syndrome, when the extra chromosome 21 material is attached to another chromosome.

## What are the effects?

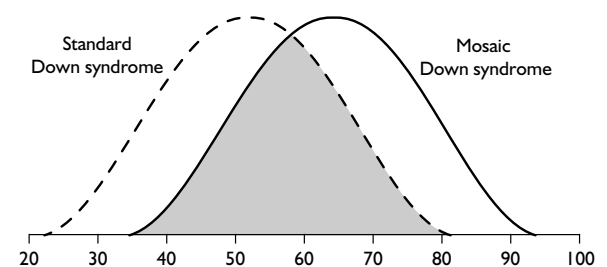
Little is known about the development of children with Mosaic Down syndrome and how it may differ from the development of children with standard Down syndrome, which makes life very difficult for their parents. There are at least two published studies that suggest that, while they have a range of health and developmental difficulties which are similar to children with standard Down syndrome, these difficulties may be less severe. One American study of 30 children<sup>[1]</sup> and one Japanese study of 8 children<sup>[2]</sup> compared them with matched children with standard Down syndrome and their findings suggest that groups of children with Mosaic Down syndrome have a higher average IQ.

However, group averages are not very helpful when the variability of children within the group is large. For example, the average IQ score for the Mosaic Down syndrome group may be 64<sup>[1]</sup> but some children may score 70 or more and some 40 or

less. The average IQ for the standard Down syndrome group may be 52,

but again some children will score 70 or more and some 40 or less. The range of scores for children with standard Down syndrome and those with Mosaic Down syndrome will, therefore overlap and some children with standard Down syndrome will score higher than some children with Mosaic Down syndrome, even if the averages indicate that the latter group may be less affected as a whole. This is illustrated in Figure 1.

***In October 2003, the first international conference on Mosaic Down Syndrome will be held in Florida, USA***



**Figure 1. An illustration of the overlap of the IQ measures for children with Mosaic and standard Down syndrome. Children with either diagnosis may be represented in the shaded area.**

These studies are rather dated and I do not think that IQ measures have much practical value, but I am using them to illustrate the issues facing parents. An individual child with Mosaic Down syndrome may show less developmental delay than a

**... if many families worldwide join the Association it will be possible to begin to gain more detailed information on the needs of these children and their families**

child with standard Down syndrome or they may not.

The findings of the first American study<sup>[1]</sup> and a more recent study of a larger group of 45 children carried out in the USA by researchers at the Medical College of Virginia, Virginia Commonwealth University,<sup>[3]</sup> can be found on the website of the International Mosaic Down Syndrome Association. The researchers at Virginia have compared the children with Mosaic Down syndrome with their brothers and sisters and also with children with standard Down syndrome.

**... very similar health and developmental difficulties for children with Mosaic Down syndrome but possibly to a lesser degree**

The progress of the children with Mosaic Down syndrome was delayed compared to their brothers and sisters on all developmental milestones except age of first rolling over, confirming that the presence of Mosaic Down syndrome can be expected to delay development in all areas. When compared with children with standard Down syndrome, there was a tendency for the children with Mosaic Down syndrome to reach all developmental milestones earlier except in the area of speech and language development.

**Some of the children do not actually look as if they have Down syndrome**

### A similar profile to a lesser degree

This study, like the earlier ones, gives the picture of very similar health and developmental difficulties for children with Mosaic Down syndrome but possibly to a lesser degree. The families have made use of the same range of services including speech and language therapy, physiotherapy and occupational therapy. All but two of the children had received early intervention services and of the 30 of school age or older, all but one

**How are these children going to feel about their identity as adults? Will they want friends to know that they have Down syndrome?**

had benefited from additional support in schools. One child was fully included without support, ten children were fully included with support and most had experienced some inclusion.

## Are individual differences linked to cell pattern?

Children with Mosaic Down syndrome can have different patterns of Mosaicism. In order to try to find out why some children are more significantly delayed than others, some researchers have tried to investigate links between the degree of disability and the percentage of trisomic cells (those with 3 copies of chromosome 21) that the child has. For example, some children have only 25% of their cells with the extra chromosome while some have 50% and some have 75% affected cells. It has been suggested that children with a lower percentage of trisomic cells, are less affected by developmental difficulties, but larger studies are needed to explore this issue, before we can be confident that there is such a relationship.

## Personal experiences and some difficult issues

We have had the experience of supporting a number of children with Mosaic Down syndrome in the past 20 years, and our evidence would support the view that these children have the same profile of speech, language, verbal short-term memory and learning difficulties but that their delays tend to be less than the children with standard Down

s y n d r o m e . Some of the children with Mosaic Down syndrome that we know do not actually look as if they have Down syndrome – the usual physical features are not obvious. This raises some important and difficult social issues and identity issues for both parents and children, which parents have discussed with us. It is likely

that no one would realise the child has Down syndrome unless told, even though the child's developmental delays may be apparent. How are these children going to feel about their identity as adults? Will they want friends to know that they have Down syndrome? Will they feel that they have much in common with other adults with standard Down syndrome? Does it help to tell teachers and other children that they have Down syndrome when they start school? Will they be treated in a more typical way if the diagnosis is not mentioned? One of the parents on the Association's website states that she decided not to tell others of the diagnosis. These are issues on which readers may have opinions that we would like to share as the implication is that society is still not fully accepting of individuals with Down syndrome and that most people would still rather not look different from the norm. Please write or e-mail if you have a view on this issue.

## The conference

If you are the parent of a child with Mosaic Down syndrome, do consider joining the Association. Details of how to join and of how to book for the conference can be found on their websites (see below). The conference is being held in Orlando, Florida so attending it could be combined with a visit to Disneyland! Dr. Colleen

**The conference is being held in Orlando, Florida so attending it could be combined with a visit to Disneyland!**



**What do we know about the needs of children with Mosaic Down syndrome and their families?**

Jackson from the Virginia research group will be speaking, as well as Dr Len Leshin, Cindy Knapp (a parent) and others. One of us will also be contributing and we hope to learn a great deal from meeting many families and individuals with Mosaic Down syndrome. We will report on what we learn in a future issue

**References**

1. Fisher, K. & Koch, R. (1991). Mental development in Down syndrome mosaicism. *American Journal of Mental Retardation*, 96, 345-351.
2. Ikeda, Y. (1973). Intellectual development of chromosomal mosaic children with Down syndrome. *Japanese Journal of Special Education*, 10, 44-59.
3. Jackson-Cook, C. (1996). *Mosaic Down Syndrome Research Study*. Department of Human Genetics, Virginia Commonwealth University, P.O. Box 980033, Richmond, VA23298-0033

**Useful sites**

www.mosaicdownsyndrome.com  
www.imdsa.com  
www.ds-health.com

**Appeal for HOT TIPS**

*Since my daughter was born with Down syndrome two and a half years ago, I have benefited many times from the tips and suggestions from those who have "been there, done it and got the T-shirt". I have heard on many occasions parents say that they received a piece of advice from another parent or professional that really made a difference, sometimes quite substantially. Some parents have invented very creative solutions to meet a particular challenge. What I have wanted to do since my daughter was born is to collect as many of these wonderful tips as I can and collate them into one source for parents. I would therefore like parents to send in tips that have proved helpful on any issue of raising child/ren with Down syndrome at any age. The tip can be very specific or general. The bottom line is that if a particular piece of information has helped you, then the chances are good that it will help someone else. I believe that parents have a wealth of experience and knowledge to share, so let's do it!*

*Please send your top tips to:*

Ann Haig  
231 Grange Road  
Rathfarnham  
Dublin 16.

*or e-mail:annhaig@iol.ie*

*Please enclose name and contact address/phone number in case of query.  
Thank you.*

**Special Needs and Families Research Project**

University of Wales, Bangor

Sponsored by the PPP Foundation

Request participation on a research project in how family members, especially parents/parental caregivers, adapt to the care of children with special needs (specifically, Down Syndrome). We are interested in

- What kind of positive experiences are there associated with the care of children with special needs, and how might these experiences help parents to adapt successfully?
- How do changes over time affect the adaptation of parental caregivers, and do various positive experiences help parents to maintain a sense of satisfaction in their caring role?

We are looking for 150 families of children with special needs in the NORTH WALES and ADJACENT areas to participate in our research project. Ideally, families will be willing to participate in our research now and also agree to be involved in a follow up data gathering phase in approximately 12 months time. The main benefits of this research relate to improving the knowledge that we have about families and how they adapt to caring for children with special needs. In particular almost nothing is known about parents' positive experiences and how these might help families to cope more effectively. Also we appreciate that participation in research takes up valuable time. Therefore, we hope that families involved in the research will feel able to accept payment for participation.

If you are interested in taking part please contact Alexandra Beck at the University of Wales Bangor, Bangor, School of Psychology, Brigantia Building, LL592AS. E-mail: specialfamilies@bangor.ac.uk Telephone 01248 388436 for more information.