

## MOSAIC DOWN SYNDROME

## An unusual profile

The role of Trisomy 21 in causing specific speech and language development is discussed by Paoloni-Giacobino and colleagues in a recent report of the development of a 14 year old girl with mosaic Down syndrome.

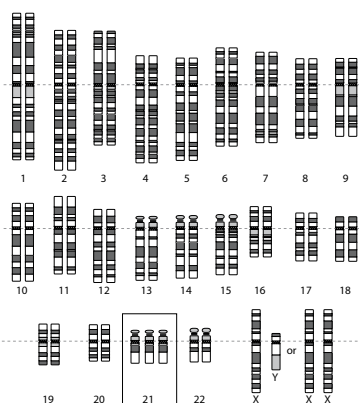
Trisomic cells are reported as being from 0.7% – 12% in different cell lines. This young girl is reported to have moderately delayed articulation and phonology and reduced fluency in her speech (affecting speech clarity and intelligibility). She also has significant delays in grammar but no impairment in her pragmatic (use of language) skills or auditory memory skills. She has an IQ score of 95 indicating that she has age-appropriate general mental abilities.

Her only difficulties are in the speech and language domain, leading the authors to suggest a direct dosage effect of a gene or genes on chromosome 21 on speech and language skills – not one that is mediated by general cognitive impairments.

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## Original research paper

Paoloni-Giacobino A, Lemieux N, Lemyre E, Lespinasse J. Specific language impairment as the prominent feature in a patient with a low-level trisomy 21 mosaicism. *Journal of Intellectual Disability Research*. 2007;51(5):401-405.



Individuals with Mosaic Down syndrome have an extra chromosome in some, but not all of their cells. The proportion of trisomic cells vary among individuals and different tissues. People with Mosaic Down syndrome can be less affected than people with full Trisomy 21.

## FAMILIES

## Brothers and sisters and Down syndrome

The literature reporting on behavioural issues and adjustment in brothers and sisters of a child with Down syndrome provides conflicting findings – some studies reporting no effects and some reporting negative effects. Difference in measures and design may explain some of the contradictory findings.

In a recent paper, Monica Cuskelly and Pat Gunn acknowledge some of the possible shortcomings of earlier studies. This study is designed to address some of them by careful case-by-case matching rather than group matching, consideration of adjustment as a multifaceted construct incorporating behavioural and affective functioning and by collecting self-reports from brothers and sisters as well as parents' reports on their behaviour.

They succeeded in studying 53 families with children with Down syndrome and 53 carefully matched comparison families. The siblings studied in the two groups are matched case by case on age, gender and birth order. The families were matched on number of children and father's occupation. They collected questionnaire information from both parents on the sibling's behaviour and from the siblings on their own behaviour and on their own self-perceptions of their social acceptance, self-worth and competencies. They collected information on daily activities and chores from both parents and children by telephone interviews.

They found no significant differences between the siblings of the children with Down syndrome and the comparison siblings on the measures of behaviour or competence. They also found no differences in the children's own reports of their relationships with friends or with academic performance. This is a reassuring finding for parents with young children with Down syndrome. On the measures they took, life for the brothers and sisters of children with Down syndrome was the same as for brothers and sisters in families without a disabled child.

There were some interesting associations between sibling self-perceptions and parent ratings of problem behaviour which applied to all the families studied. For



**A recent study finds no significant differences between the siblings of the children with Down syndrome and a carefully matched comparison group on a wide range of measures.**

example, parents reported fewer externalising (e.g. acting out, aggressive behaviours) behaviours for siblings who were more satisfied with their own behaviour and mothers reported fewer internalising (e.g. anxiety, depression) problems for siblings with more positive self-perceptions of social acceptance. There was also a link between household responsibilities and adjustment with mothers reporting more externalising behaviours when girls reported more chores and more internalising behaviours when boys contributed more caregiving. There were no group differences in the number of chores reported but the children reported more chores than their mothers acknowledged. These, and other findings explored in the paper, make this an interesting study of sibling behaviours and parent versus child perceptions as they apply in all families.

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## Original research paper

Cuskelly M, Gunn P. Adjustment of children who have a sibling with Down syndrome: perspectives of mothers, fathers and children. *Journal of Intellectual Disability Research*. 2007;50(12):917-925.