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LET'S CELEBRATE DOWN SYNDROME

Does that sound bizarre? Then let's hear why this condition doesn't need to be a downer

BY MARGOT BERTELSMANN

World Down syndrome day is recognised as a global awareness day by the UN – and it falls on 21 March for a reason. More than 90 percent of people with Down syndrome have the trisomy 21 version, which means that they have three (full or partial) copies of the 21st chromosome in the nucleus of each of their cells, instead of the usual two. March, the third month, references the three in trisomy, and the 21 references the 21st chromosome. In honour of the day, let's learn some more about the syndrome, the people it affects, and their families.

READ MORE
 Probably the world's most popular mom blogger on Down syndrome, Kelle Hampton writes a fantastic blog, *Enjoying The Small Things*, at www.kellehampton.com

THE FACTS ABOUT D.S.

People with Down syndrome present with learning disabilities ranging from mild to moderate, distinctive facial features such as a protruding tongue, flat nose and slanted eyes, a short neck, and sometimes with short stature, increased flexibility, short fingers and certain heart conditions. Down syndrome is congenital – it happens during the cell division process in the very early stages of pregnancy. In most cases there are no known inherited or environmental causes, although it occurs more commonly in older women's children.

According to the US-based National Down Syndrome Society, Down syndrome is the most common genetic condition in the world. Down Syndrome South Africa (DSSA) on its website, www.downsyndrome.org.za, cites 2008 estimates from the US infant health organisation March of Dimes that one in 500 births in SA is "affected by" Down syndrome. "This is a count of live births, although sadly, especially in less resourced parts of South Africa, many children with Down syndrome die within their first year of life," says Vanessa dos Santos, director of Down

Syndrome South Africa. "Many South Africans don't have access to regular antenatal testing, so while fewer parents therefore face the decision whether to terminate a pregnancy with a Down syndrome diagnosis, many diagnoses are also missed, and children diagnosed only later in life."

WHAT THAT REALLY MEANS

We've come a long way, in some ways, since the condition formerly known as Down's syndrome was first described by Dr John Langdon Down in 1866 (the possessive was dropped in the US to remove the sense that

the condition belonged to Down or the misunderstanding that Down actually had the syndrome). Organisations such as Down Syndrome South Africa and international equivalents exist to advocate for people with Down syndrome, and our South African legislative framework prohibits discrimination against people with disabilities – at least in theory.

But Down syndrome activists know that ever better antenatal screening internationally means that many more expectant parents know of the diagnosis before birth. And there are

two reasons a parent would want to know: to prepare for raising a child with Down syndrome, or to terminate the pregnancy. Clearly the concept that their condition is one that should, in an ideal world, be eradicated is hurtful to people who live with the syndrome.

Vanessa stresses, however, that DSSA does not consider a diagnosis with the condition alone as a reason to terminate a pregnancy, although she's respectful of every parent's choice to terminate or not. Termination of an "abnormal" pregnancy is legal in SA. Every parent needs to make their decision according to their unique set of circumstances, abilities and resources.

DSSA advocates for inclusivity and equal access to life's opportunities for people with Down syndrome. This includes mainstreamed education for many children with Down syndrome. ►



talking point

"Our schools are not allowed to discriminate against students with disabilities," says Vanessa. "Like a blind student should receive materials in Braille and a hearing impaired student needs hearing aids, a Down syndrome student may need a facilitator, which is something the government should provide (but usually doesn't). It's no surprise that many already stretched schools resist accepting disabled students – but DSSA is prepared to advocate for individuals and parents in addition to its role as a DS advocate at national and legislative level."

After school there is "a gaping need" for employment, says Vanessa. "We advocate for open or supported employment for people with Down syndrome in the open labour market. But there is a very long way to go."

MEET THE FAMILY

Lauren and Alec Schoeman have three sons: Adam (six), Ryan (four) and Nicholas (almost two). Nicholas has Down syndrome.

Q NICHOLAS' CONDITION WAS NOT DIAGNOSED ANTENATALLY...

Nicholas is our third son and my previous two pregnancies had gone smoothly. I was 32 when I fell pregnant, my 12 week DS blood screening came back negative and Nicholas's nuchal fold was within normal range. There was no reason to suspect Down syndrome.

Q HOW WOULD KNOWING HIS DIAGNOSIS ANTENATALLY HAVE CHANGED THINGS FOR YOU?

We naturally would have been devastated. It's a huge shock for parents to hear the news at any stage, be it in utero or at birth. Suddenly the expectations you had for your child change and you're following a different path. Although we didn't have time to prepare, in a way we're relieved we didn't know. Choosing whether to terminate would have been heartbreaking and agonising. Our choice was made for us and I'm so



The Schoeman family can't imagine life without Nicholas

grateful for that. I can't imagine a life without Nicholas and we couldn't love him more if we tried.

Q WHAT HAS BEEN THE HARDEST PART OF COPING WITH NICHOLAS'S CONDITION?

Society's limitations and prejudices placed on Nicholas, purely due to his extra chromosome. Nicholas is who he is and I hope that we can help him reach his full potential.

Q WHAT HAS BEEN EASIER THAN EXPECTED?

Nicholas integrated into the family so naturally. From day one his two brothers were besotted with him and he lights up when they're around. At four months Nicholas had open heart surgery to repair a hole in his heart (a common occurrence in Down syndrome babies). It was a traumatic



time for our family, but a year later his heart is perfectly healthy. Our lives were initially turned upside down, but we now carry on like any other family. That has been the greatest surprise. Life is indeed busy, but that's par for the course when you have three small children!

Q THERE ARE MANY STEREOTYPES WHEN IT COMES TO D.S. – "STUPID" AND "HAPPY" COME TO MIND. HOW DO YOU ENCOURAGE OTHERS TO HAVE A MORE

NUANCED RELATIONSHIP WITH NICHOLAS, AND WITH DOWN SYNDROME IN GENERAL?

Nicholas can't be cured of his intellectual disability, but there is so much more to a person than their IQ. He has already changed people's perceptions of what it means to be "normal". I often hear that people with Down syndrome are "always smiling and happy". This is not the case. They are people first, each with their own unique characteristics. If they are always happy, then that's because of their nature as a person. That's not due to Down syndrome. Nicholas hasn't been a great teether, so when he's cutting a new tooth, he's miserable just like most babies. His emotions are just like the rest of ours. And we're getting to know his stubborn side at the moment!

Q ANY ADVICE TO PARENTS WHO HAVE JUST RECEIVED A DOWN

SYNDROME DIAGNOSIS?

Remember that your little baby is a baby first. It's YOUR baby. Down syndrome will not and must not define them. I also suggest speaking to parents who have Down syndrome children. It helped me so much and I, in turn, hope to assist other families. Yes, life will be different to what you had expected, but different is not necessarily a bad thing. I think you'd search far and wide to find a parent who regrets having that child and where that child doesn't bring joy to the family. The most important thing is to seek knowledge and make an informed decision, not one out of fear or ignorance.

We're learning every day about Down syndrome having Nicholas in our lives and we welcome open and honest communication, be it with friends, family or strangers. We appreciate people taking the time to care and wanting to be educated. It's all about creating

awareness and being equipped with the necessary knowledge, especially if you are faced with life-changing decisions.

Q WHAT ARE YOUR HOPES FOR NICHOLAS' FUTURE?

Our hopes for Nicholas are good health, happiness, to be constantly aware of the love we have for him and to be given the opportunity to follow his dreams. Down syndrome is not a death sentence, nor a disease. These beautiful children can lead long, happy lives and if given the chance, can be a real asset to society. Nicholas has brought so much joy to our family and we are so privileged to call him our son and brother. I know that he is going to make a positive difference in people's lives and most importantly, he already has in ours. **YB**

Lauren wishes to encourage parents who need support to contact her on laurenschoeman@telkomsa.net