

The Down Syndrome Research Foundation UK

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Dear Supporters of the DSRF,

We all learn how to survive as babies and smiling gets a smile as a reward. This survival instinct is something we inherit from our parents, and 100% of our genes are from our parents.

A child with Trisomy 21 is entirely like its parents. Not more like other children with Trisomy 21, (not a new race of people). They may look similar but genetically every child is unique and very different. They develop a personality that is a response to their environment and the support they get from their family and from those around them. Faced with challenges the parents teach them to be courageous. Their personality is not a genetic accident it is something they created as a response to challenges. Medical research is not going to change that personality. Those extra genes are also from the parents and they are making chemicals which disturb the precise functioning of the other genes. This is especially true in the brain where signaling is controlled by chemicals made by the genes.

We can now understand what is going wrong with this signaling and it has nothing to do with a happy smiling face or jumping for joy. It is entirely to do with parts of the brain being shut down by chemicals that are disrupted by those extra Trisomy 21 genes. Medicine to help the brain should not scare anyone. If we live long enough we will all need that medicine. Curing the mental disability caused by Trisomy 21 is really a search for a very safe (targeted) treatment therapy to allow proper brain function where we know what chemicals are over expressed. Most likely it will be a pill that is taken every day and something that wears off in a couple of weeks if it is not taken. There is evidence of brain injury that leads to dementia and to Alzheimer's in the adult years. Any therapy to improve memory will include a therapy to reduce this very high incidence of injury to the brain. We have to do this research – it is very important and nothing to be afraid of.

Remember: That wonderful personality is not a genetic accident; it is something you created together and it is there for life.

Have a Merry Christmas and a Happy New Year.

Peter Elliot

A letter to the BMJ

The British Medical Journal (BMJ) recently published a paper on screening and abortion of babies with DS. This was our response to them and was published on their website:

“If 94% of parents are electing to have an abortion when they learn the baby has Trisomy 21, then these doctors must be impressing the parents with a very biased, fearful and distorted scenario of what it means to have a child with Down syndrome. What the future holds for this child is something these doctors can’t know anything about, and yet they are frightening these parents and telling them to get an abortion.

The parents with this diagnosis will not know anything about Down syndrome. But they can be frightened into getting an abortion which is a huge decision that requires a lot of pressure from their doctor. So, this is not an informed choice by the parents. These advocates of abortion have a hidden agenda that seeks genetic perfection, driven by a government objective to reduce healthcare and special education costs for the nation.

Here is the real information for new parents:
http://www.dsrf-uk.org/PDF/BrightBeginnings_3.pdf

Apart from the fact that screening and abortions are immoral. The strength of the human race lies in our genetic diversity. Killing the weaker members of society is inhumane and it is not making us better or more perfect human beings when we do this. It degrades all of society.

Modern medicine can cure or treat most of the developmental and medical problems faced by these children. The last remaining challenge is the learning disability, and this will also be cured in the near future.

The current evidence would indicate this research is very feasible. Short term memory is impaired in people with Down syndrome and we can study the brain chemistry and investigate the differences in this brain. This can be the key to improving cognition and all levels of mental ability in this population.

Drugs are available which can be tested but first we need to investigate brain function in this population.

A cure for the learning disability is a more than a possibility it is within our abilities. We have the technology and the new medications to get this research underway.

Doing a body count of the abortions is not research and it should be a wake up call to all researchers when they see this article in the BMJ.

I have a vested interest in research because I have a 24 year old son who has Down syndrome. He is very healthy and he is an important member of our family. He makes all of our family better and less selfish individuals and he teaches us how to behave and care for each other.

I founded the Down Syndrome Research Foundation in the UK in 1996. This is a registered charity. We are initiating research to improve brain function in patients who have Down syndrome. Our website is www.dsrf-uk.org."

Peter Elliott
Research Director and Chief Financial Officer

This is an article in Discovery Magazine,

This is not a cure for the disturbed chemistry that increases the risk of dementia and Alzheimer's. Research is underway and planned in this area as well. There is a need to cure the brain function problems and the brain injury problems at the same time. A medical therapy that works will improve the lives of these children, allowing them to have a more normal life and reach their full mental potential. – Peter Elliott

Drugs Could Reverse Down Syndrome Symptoms

A newly-found neurotransmitter link to the condition's associated learning disabilities could be targeted.

By Josh Clark | Mon Nov 23, 2009 06:30 AM ET

The learning impairment that characterizes Down syndrome and keeps people with the developmental condition from adapting to new environments could be reversed with a simple drug regimen, a new study suggests.

The Stanford University study, led by physician and neuroscientist Ahmad Salehi, was conducted on mice genetically engineered to develop a rodent version of Down syndrome.

In humans, the condition -- also known as trisomy 21 -- results from the presence of an extra copy of chromosome 21. (Chromosomes typically come in pairs.) The additional copy of the chromosome adds between 130 and 300 extra genes, which are responsible for characteristics of Down syndrome-like cognitive impairment.

Cognitive impairment in Down syndrome is the result of a breakdown in the function of the hippocampus. This region of the brain is responsible for contextual learning, or gaining and applying knowledge in real-world situations. Recalling the location of certain items is a prime example.

"Remembering where your keys are isn't just based on navigation," Salehi told Discovery News. "It also includes the sound the keys made when you placed them down and all of the other little bits of information involved."

The hippocampus pulls all of these disparate sensations and awareness of our bodies in relation to our surroundings together to form new memories. When we need our keys again, we draw on these memories to help us find them.

People with Down syndrome may have trouble forming such memories, and Salehi and his team appear to have pinpointed the reason why

Salehi and his team probed the hippocampi of the mice used in the study and found that the region wasn't functioning correctly due to a lack of the neurotransmitter norepinephrine, a chemical that allows neurons in the hippocampus to form the connections that form contextual memories.

In the study, Salehi and his team found that this dysfunction could be corrected in the mice -- and surprisingly easily. The researchers injected modified mice with a drug that is converted into the norepinephrine within the brain.

Within five hours of injection, the modified mice that previously couldn't learn to fear a tone accompanied by an electric shock could now remember the conditioned response. The modified mice also learned to nest in a novel environment, which is a standard sign of intelligence in the rodents.

After being injected with the drug, L-DOPS, the mice modified to express Down syndrome showed no significant difference from their counterparts in terms of intelligence.

The study was published in the Nov. 18 issue of the journal *Science Translational Medicine*.

Salehi says he was surprised by how quickly the effects of the cognitive impairment were reversed, but he is cautious about predicting a similar strategy could work on humans. Salehi says that when the mice were tested again after two weeks, they had returned to their previous levels of intellect. He also points out that what works on mice may not work on humans. In addition, he asserts, Down syndrome is a multifaceted condition; cognitive impairment is just one of its characteristics.

Richard Urbano, a Down syndrome researcher at Vanderbilt University who didn't take part in the Stanford study, said that Salehi's findings could show real promise. Like Salehi, he is cautiously optimistic.

"If this finding held in humans, it would be a really important one," he said. "But just because it works in mice, it doesn't mean it will work in humans."

Drugs that introduce norepinephrine to the brain are already widely available, making Salehi's treatment strategy closer to a reality -- if it can be proven to work.

"If in the future it turns out that this is the right strategy, the good news is we do have access to drugs that already do this," Salehi said.

TS65DN Mouse research information: an update

Learn about how the mouse with DS was developed and the important research that has made discoveries in Cancer research, Heart research, and in Brain research.

The Reeves lab (<http://inertia.bs.jhmi.edu/>) at the John Hopkins University complements genetic analyses in human beings with the creation and characterization of mouse models to understand why and how gene dosage imbalance disrupts development in DS.

Watch this NDSS webcast video by Dr Roger Reeves:

<http://tinyurl.com/reeveslabvideo>

It is an hour long presentation. The first 20 minutes is a review on the development of this mouse that has characteristics of human Trisomy 21.

The last 40 minutes is a review of discoveries using this mouse which can be directly related to humans with Down syndrome, including brain research.

Spare an hour to watch the video. Let us know what you think.



It's time to swing! **Erin Dine** has some fun.

Shelby goes to school

by Suzanne McCloud

I have to pinch myself! Our beautiful daughter, Shelby, is starting school in January. Where did that time go? It seem like only yesterday that we gave birth to our fourth child in the very early hours of 8th August 2005, and minutes later, it seemed like her birth was surrounded by a big black cloud. How wrong we were!

She has brought us so much laughter, tears and most importantly **taught** us so many things.



Shelby has been attending a mainstream preschool for the last 2 years. We have gone from being worried about her drinking from a cup to now going to the toilet, pretty much unaided. All our worries were wasted time! She is able to read several words, knows all her colours, shapes and can count to 5, has started talking, loves anything that involves climbing, dancing and singing.

We have all been gently guiding her to this moment - starting school. With lots of support from her keyworker Donna, her pre-school teacher, Jo and support from the school she is going to attend, Shelby has begun her transition.

She started going to school one morning a week initially, building it up to 3 sessions before she starts in January. She has done PE and loves it, encouraging her classmates to go under the bench rather than over!

Her classmates are all learning to sign, and so are the rest of the school, they all seem to be sponging the information in, desperate to learn the next sign. Every morning when we go and line up for the start of the day, kids come bounding up to her and sign "Good morning". Shelby, taking it all in her stride, just says "Hello". She positively revels in all the attention she is receiving at the moment.

She recognises her teacher, the headmaster, and several of her peers, by name, and asks me everyday if she is going to school. She loves putting her school uniform on and is totally independent with getting undressed and dressed and by all accounts is quicker than her peers at doing this.





I am sure we will have our moments. I fear that her Statement hours are not enough. I worry with Shelby being the first child with DS at the school that they are not going to have the correct resources and teaching strategies. Will they understand Shelby's imperfect signs and sometimes unclear speech?

I am having to let go and am not going to be able to be there and as involved as closely as I have been to this point. This has caused me many hours of tears, but it is part of her growing up, growing independent and more importantly, letting Shelby become the beautiful person she is destined to be.



Harvey Simpson entered the Next Generation Children's competition (organised by NEXT UK to find models for their catalogue) and made it to the top 100. He rested at 12 out of 20,000 children.

Here, his proud mum, Lisa, tells us about this journey.



“ My inspiration for entering Harvey came from a post seen on Blair Williamson's site about a little girl with Down Syndrome in America that was modeling for an American catalogue.

It was an intense three weeks of constantly being on the computer and pestering people to vote and pass on the link.

Due the voting process two local newspapers ran with the story and we went on local BBC radio Derby.

The whole reasoning for me putting Harvey in this competition was my desire to see change. You never see our kids with Down syndrome used in fashion catalogues. I want people to see that **you don't have to be afraid of Downs Syndrome**. I want people to see my little man who not only has Downs Syndrome but has overcome open heart surgery, that it doesn't matter. He runs, plays, acts, loves, cares, just like any other and that Downs Syndrome is NOT as reason to terminate a pregnancy.

Through all of this, if I could just help to stop just a hand full of parents from terminating then I would be happy.

Times need to change. Our children and adults with Downs syndrome are part of our society and so should be represented in society properly i.e.: being used as models in catalogues. The more our kids are represented in everyday society the more likely fear will be dispelled.

Anyway, **we are now in the final** and going to Woburn Safari park on the 21st of December for the final and a big Xmas party!!

Fingers crossed for change!! ”

- Lisa Simpson

Your Christmas Gift to us and ours to you.

Christmas is almost here. This year, please consider supporting our **Progress Appeal**.

Why we need your support

There are many children, youths and adults who live with Down's syndrome and as such, many have a severe learning difficulty. Sadly, the condition of Down's syndrome often carries with it, a serious decline in health involving heart disease, diabetes and leukemia as well as increased memory loss as they reach young adulthood. Premature dementia and Alzheimer's can often set in at the heartbreakingly early age of mid to late 30's.

Our work here at the DSRF (UK) is to research ways to slow down and potentially delay the onset of these conditions. But to do that, we need to raise funds to enable us to commission these vital studies and find ways to help improve the quality of life for the many children and adults who have Down's syndrome – and those who will be born in the future.

This is why we have created **the Progress Appeal**. Our work here at the DSRF is so vital and now, more than ever, we depend heavily on voluntary contributions.

So you can see the impact that fundraising for the DSRF will have. Every kind gesture, every small collection or personal cheque will add up and enable us to move our research forward and get a step closer to finding the answer.

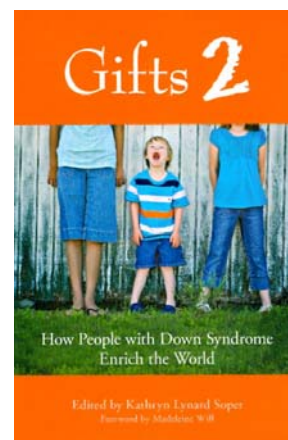
Fundraising and volunteering can be an exciting rewarding way to support us and we want to help you make the most of every moment. There are so many ways that you

can get involved, so please read on and explore our fundraising and event pages and take a look at our downloadable DSRF (UK) Fundraising Pack (on our website) which just might make your challenge easier.

For news on our fundraising events, ideas and opportunities please refer to the menu to the right of this page or contact Fundraising@dsrf-uk.org

If you donate £50 or more towards our Progress Appeal this Christmas, we will send you a free copy of this wonderful book 'Gifts 2: How people with Down Syndrome enrich the world' (RRP:£16.99).

"Acceptance, courage, friendship, awareness, and joy - these are the gifts recounted in more than seventy inspirational essays included in "Gifts 2".



If you choose not to (or cannot) contribute to our Progress Appeal at this time, why not instead buy this book as a Christmas present for a family member or friend? Please email peter.elliott@dsrf-uk.org for more information or to place an order.

DLA – saved for some: an update

Many of you may have been following the developments since the Guardian first reported (in July 2009) that the Government is planning to axe the Disability Living Allowance (DLA). This announcement was met with a lot of protest and as a result, there has been some softening in the government's stance recently.

Here's an update from the Benefits and Work website (<http://www.benefitsandwork.co.uk>)

DLA saved – for some 27 October 2009

It's a start, but nowhere near enough.

Health secretary Andy Burnham has said that he has 'heard the concerns and worries about disability living allowance'. As a result, he has announced that:

"I can state categorically that we have now ruled out any suggestion that DLA for under-65s will be brought into the new National Care Service."

Good news indeed . . . for some . . . for the moment.

But definitely not for the one and a half million people who depend on AA.

Nor for the for the three quarters of a million people aged 65 and over who receive DLA.

Not even for the 400,000 DLA claimants currently aged between 60 and 64, many of whom will have reached the age of 65 by the time labour's proposed National Care Service is introduced.

Because, of course, DLA is not just paid to people under 65. You have to make your claim before you are 65, but you can then go on claiming indefinitely if your needs do not change.

Unfortunately, many organisations who should know better seem to have forgotten that – perhaps just as the government hoped.

Because Mr Burnham made no secret about why he made this announcement: he wants to shut people up. He said in his speech, given at a conference in Harrogate on 22nd October and also published on the Big Care Debate website:

"One avenue I do want to close down, however, is the debate and controversy over Disability Living Allowance."

In that ambition, he seems to have succeeded, at least so far as some disability charities are concerned.

Immediately following Burnham's speech, Disability Alliance sent out a press release stating that:

". . . the Disability Living Allowance (DLA) benefit will not be affected by Government plans to merge some benefits with social care funding . . . Andy Burnham's announcement will reassure disabled people that DLA is safe – for now at least."

The Disability Charities Consortium told the media:

"This represents a real victory for disabled people who felt very strongly that the DLA should be retained and made their collective voice heard on this issue. "

Macmillan Cancer Support also issued a press release saying that:

"Whilst we are pleased the Government has said Disability Living Allowance (DLA) will not be used to meet the shortfall in social care

funding, we remain deeply concerned that Attendance Allowance (AA) is still under threat.”

But that isn't what Andy Burnham said at all. He said DLA for under 65's is not being considered.

This was echoed by Yvette Cooper, the DWP secretary of state who told a meeting of the All Party Parliamentary Group on ME on 21st October that DLA for people of 'working age' is not under review.

It was also made clear by Burnham that there will be no transitional protection of existing awards for current claimants. Instead, 'an equivalent level of support' will be provided by your local authority.

Burnham's announcement seems to have had the desired effect, however – the 'debate and controversy' over DLA appears to be over as far as some disability charities are concerned. Yet, in a little over two weeks time the deadline for submissions on the green paper ends.

It's vital that the case for saving DLA for all claimants is still made. Only now there is a real worry that not only have the disability charities relaxed, but also that Burnham will claim that because 3,000 submissions to the Big Care Debate were made before his announcement that DLA for under 65s is safe, they should mostly be discounted.

If you don't want the government to get away with closing down 'the debate and controversy over Disability Living Allowance' there are things you can do.

Contact disability groups you have a connection with and warn them that they still

need to respond to the green paper in relation to both DLA and AA.

Respond to the Care Green paper yourself, again if necessary, making it clear that you are aware that DLA for under 65s is not under consideration and giving your views on axing AA and DLA for people aged 65 and over.

<http://careandsupport.direct.gov.uk/greenpaper/execsum/>

Email: careandsupport@dh.gsi.gov.uk This e-mail address is being protected from spambots. You need JavaScript enabled to view it

Rouse people to sign the No 10 petition, which is gathering real momentum again: it now has over 19,000 signatures and is at number 8 out of over four and a half thousand petitions on the site. Not bad going for a petition that has been running for less than two months.

<http://petitions.number10.gov.uk/AttendanceA/>

Tell your MP what you think or, better still, go and visit them and tell them face-to-face.

One final thought. The revelation that the government is considering slashing the income of 2.5 million older disabled claimants was made by Andy Burnham in a keynote speech last week.

The subject of that speech?

Outlawing ageism in the NHS.

Good luck,

Steve Donnison

Read more here: <http://www.benefitsandwork.co.uk/news/latest-news>