**The All-party Parliamentary Group**
**for 22q11 Syndrome**
**Launch**

**Parliamentary Reception**

**4-6pm on Tuesday 10th March 2015,**

**House of Commons**
**London**
**SW1A 0AA**

# Itinerary

**4pm** **Arrival and tea**

**4.30-5pm:** **Presentations**

**Jack Lopresti MP.**

Chair of the APPG.
Conservative member for Filton and Bradley Stoke.

**Jane Ellison MP.**

Conservative member for Battersea
and Parliamentary Under-Secretary (Department of Health).

**Dr Alex Habel. 5 mins.**
Retired Paediatrician, Great Ormond Street Hospital.

**Dorne & Lee Mitchell**
mother and father of Ivy.

**Julie Wootton.**
Chair of trustees Max Appeal

**5-6pm** **Guests mingle**

## Purpose of the event

To raise awareness of the diverse needs of children, teens and young adults with 22q11 DS – a complex condition that is often undiagnosed and leads to lifelong struggles for those affected. The syndrome is estimated to effect up to 35,000 people in the UK.

These conditions affect health and quality of life from birth through infancy and childhood into adult life with over 180 physical, functional and psychological associations reported. The complexity of 22q11 frequently leads to clinical confusion and a delay in diagnosis often by years.

The event is being hosted by Jack Lopresti, MP who recently set up an APPG to raise awareness of this medical condition after meeting one of his constituents who has a daughter with the condition. The other officers of the group are Nigel Adams MP, Paul Uppal MP and long-time supporter of Max Appeal, Margot James MP.

## Materials for the event

Four posters highlighting some families who are members of the charity Max Appeal and showing the growth charts specific to 22q11DS. Thank you to the following families for offering their support:

- Ivy, 2yrs, and her mother Dorne.
- Harley, 13yrs and his mum Deb.
- Hannah, 23yrs, and her mother Debbie.

# Hannah's story: told by her mother Debbie

***"We had spent years hitting brick walls trying to get support for Hannah – we were at the point where we were literally begging for help. One day in August Hannah decided she could no longer cope– she began to see black bits, couldn't swallow which led to voices telling her jump of our kitchen roof trying to end her life."***

From a very young age Hannah suffered from high anxiety and we had a lot of difficulty controlling this along with her challenging behaviour.

After years of searching for the right kind of support, we decided to sign up for a research programme in Geneva when Hannah was 9yrs. On our first visit they diagnosed depression AHDD, Autism along with a host of other difficulties. This was a lot to take in but we were hopeful that Hannah would get the support she needed back at home.

On our return to the UK our first hurdle was to fight to get her seen at child development clinic by Psychologist. I'll never forget that during that first nerve-wracking appointment she took a call from an estate agent as she was buying a house. Her focus certainly seemed somewhere else. She then proceeded to ask what *we* wanted her to do. She prescribed Lamotrogin for mood swings and we were left to it - no therapy.

During Hannah's early teens we found ourselves fighting to get her seen by a consultant psychiatrist who told us he didn't feel qualified to see her. At that point we fought for funding to be seen at the Maudsley Hospital.

Following our appointment a comprehensive report was sent to our local team with a range of recommendations. Sadly, much of this was ignored and instead it was de-coded and interpreted that we needed family therapy. Our family wasn't broken we just needed help for Hannah! During these therapy sessions they never once spoke to Hannah but insisted our other two daughters attend.

This whole process blew our family apart. It took us many, many years to repair the damage with our other daughters. As well as the damage to our family, Hannah had been totally let down by the system. It was utterly heart-breaking for us all as family.

At around the age of 17yrs Hannah continued to deteriorate. Our GP referred us to adult services for learning disabled patients. During all these years we kept asking the different teams to liaise with Geneva, who we had been seeing on a three year basis. The team in Geneva could quite clearly see the help Hannah required but no one here would contact them. It was so frustrating.

We sensed we were entering a dangerous phase and Hannah was clearly reaching tipping point. During her 19th year she rapidly got worse. We were asking many questions and she was writing down her different experiences; voices, hallucinations and indeed some physical difficulties. We questioned on many occasions if it was the drugs they refused to consider this or investigate if there was a physical cause either.

In the six months leading up to Hannah's attempted suicide our medical professionals insisted on increasing the Risperidone. We were literally begging them to contact Geneva as they were suggesting that this drug. They refused to listen to us

In early August that year they prescribed an anti-depressant alongside her anti-psychotic drugs which they continued to increase. I kept on asking about the drugs and alerting them to the fact that she had a pre-existing heart condition. They assured me it was fine.

During this time Hannah told me she saw black bits and couldn't swallow. This eventually led to voices telling her jump off our kitchen roof trying to end her life. She couldn't cope.

The day before we took Hannah to A&E with our deep concerns – she was told to go home, smile and have a nice weekend. The next morning we had an emergency appointment with the Psychiatrist where we literally begged them to help her. He refused to believe it was anything but her head and told us to take her home as he would not want his daughter in a ward like theirs.

That afternoon it happened.

Three weeks into her admission at St Marys for her acute injuries the Psychiatrist came to visit. He insisted on speaking to her on his own. He wanted to admit her to a psychiatric ward on discharge but they would not be able to look after her physical injuries from the fall. St Marys Hospital refused this and, with support, she came home to be nursed by us - much to our relief.

Hannah still continued to see black bits and have difficulty swallowing. In early 2012 we saw two people; my husband asked his cardiologist to see Hannah no matter what the cost. He was wonderful saw her on the NHS within a week and diagnosed a serious arrhythmia problem inflamed by all the drugs she was taking. We also saw an ear nose and throat consultant.

Our psychiatrist phoned before our appointment and told him she had Schizophrenia - this has never been diagnosed. After some persuading a camera was put down her throat and diagnosed a severe form of Laropharyngeal Acid Reflux.

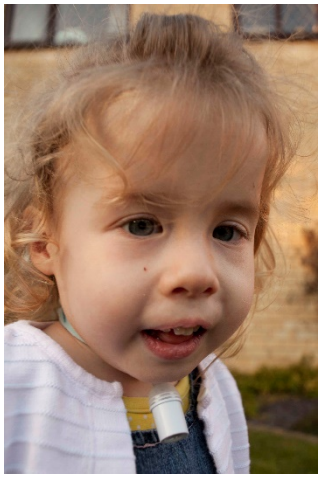
At this point we raised a formal with our mental health authority who admitted that failings had taken place. Not surprisingly we have a total lack of trust in the system that was supposed to help us and Hannah.

Today, Hannah's care remains patchy. We've just had had another change of Psychiatrist. We recently went to see our local MP - he was dismayed at Hannah's treatment and wrote to the head of Central North West London Health Authority, who in turn wrote to the lead clinician for services with people who have learning difficulties. He has requested our new psychiatrist to hold an urgent meeting with ourselves.

I feel that we are finally being listened – literally in the last few weeks. A case hypnosis has been written on Hannah's care and some serious failings have been highlighted and agreement that her medication should have been changed.

We now hope to move forward with a comprehensive care plan.

## Ivy's story: told by her mother, Dorne



We were over the moon in 2011 when we discovered we were expecting our second child. As we approached the last week of our pregnancy we received the bombshell that our little girl appeared to have abnormalities. A test confirmed that Ivy had Di-George Syndrome – a condition we had never heard of – along with that came the devastating news that she had a rare and complex cardiac problem which would see her require open heart surgery throughout her life.

Three weeks after receiving the news I was induced. We welcomed the most beautiful and perfect looking baby into the world on 6th August 2012. We, along with the medics, were amazed at how well she seemed to be coping despite her internal abnormalities. We took her home 12 days later.

Little did we know of what was to come. At 3 months Ivy caught a cold just like other children at that time of year, but to a child-like Ivy who is immunosuppressed it knocked her for six. She was urgently admitted to hospital and put on life-support.

Ivy failed to come off ventilation and after many weeks of emotional highs and lows, we were told the bronchiomalacia she had was extremely severe - her pulmonary arteries were compressing her airways. Ivy was very sick in severe heart failure. Our beautiful baby had become unrecognizable.

The day after Boxing Day she underwent open-heart surgery. This was the longest day of our lives. After 9 hours we received the call that she was successfully out of theatre. A few weeks later she underwent more investigations as despite several attempts she was still unable to breathe alone.
The damage her heart had caused to her airways meant she needed long term ventilation and a tracheostomy. Hopefully this would stop the episodes of her going blue – well, almost black - as she was revived daily. We were told to expect the worst on several occasions

Since then she has had three more surgeries - a repair to her diaphragm, her tracheostomy and a gastrostomy. . In July 2013, after 9 months on intensive care, Ivy finally came home. We were over the moon. We were always determined she would be home for her 1st birthday - that was our focus and we never gave up on it.

The months in hospital saw our stress levels go through the roof; the constant daily 3 hour plus commute, trying to maintain some normality for our eldest daughter, trying not to drown in worry and fear, it was almost unbearable at times.

Despite all this, Ivy continues to amaze the doctors with her strength and determination to survive. Her battle is far from over, we don't know what the future will hold, nor do we want to think about it, but some days I can't help myself and I cry. We hope Ivy will grow old but sadly she may not. We are full of pride for our little Ives. She has changed our lives in so many ways – she puts life into perspective.

Today, thankfully, we are delighted that Ivy is making remarkable progress. She is spending time off the ventilator, is running around, beginning to sign, and remained in relatively good health despite her immunity issues. Her one kidney is functioning well and her cardiologist keeps a close eye on her heart. We just have to watch and wait for the next open heart surgery. Each surgery brings new fears for Ivy and us, it gets harder that's for sure.

Ivy is the most adorable, cheeky, spirited little girl. It's her grit and determination that sees her still with us today, along with some of the best doctors and surgeons we could ask for.

# Harley's story: told by his mother Deb

The moment Harley was born it was clear to me that something wasn't quite right. He was lethargic, looked 'elf like' and wouldn't feed; he just didn't thrive – I was so worried but none of the medical professionals seemed eager to find out why my son was so small and frail.

I'll always remember taking Harley to the dentist. His teeth's enamel was weak and patchy, his milk teeth looked awful - despite me cleaning them daily and vigilantly keeping an eye on his sugar intake. The dentist insisted that teeth only get this way with too much sugar and poor brushing. Poor calcium levels is common in 22q, which explained Harley's milk teeth.

Harley's speech was also a concern. He had a nasal tone and couldn't explain his thoughts in a methodical manner with a set time frame. As a toddler he was a real livewire. He never stopped, never wanted to sleep, climbed on and up everything and threw anything in sight. You had to watch him like a hawk. In pre-school in was a different story, he was subdued and totally overwhelmed - like a child lost in a confusing world.

As he reached the first year of primary school his behaviour rapidly deteriorated. He just couldn't function in the classroom. I pleaded with my doctor and other professional agencies to assess if he had ADHD. His condition also impacted on his sisters, now 24yrs and 21yrs. Harley was challenging, argumentative and had no regard for their possessions.

Eventually various tests were done for suspected syndromes, all coming back negative. Harley had low levels of vitamin D and was given a supplement and a paediatrician noted that Harley had 'mild facial dysmorphia'.

I am certain that if awareness of 22q and its many symptoms was higher, all these symptoms would have been put together and a 22q diagnosis would have been made at this time. So far we had all the symptoms; slow growth, patchy tooth enamel, nasal and delayed speech, behavioural problems, mild facial dysmorphia, low vitamin D levels – the list goes on but the medical professionals were baffled. Looking back *I think this is so very significant as 22q is the second most common syndrome after Downs Syndrome yet so rarely diagnosed.*

It took years and years of fighting to find the answer. We were finally referred to Geneticist and a simple blood test gave us our diagnosis. It was the missing piece of the jigsaw for me, mystery solved.

The diagnosis floored Harley, he was extremely upset about being born with this condition - it was the 'why me' scenario. He was diagnosed the summer before starting secondary school. A huge transition in itself - Harley started feeling angry and resentful, which showed in his behaviour.

With a diagnosis I was optimistic that he would have a level of understanding from his schools and intervention would follow. How wrong I was! No reasonable adjustments were made for Harley and he faced outright disability discrimination on several occasions from being excluded from the last residential school trip which included a 'far-fetched' risk assessment to our absolute lowest point when the school outright refused to educate him for a month. I was entrenched in a long political battle with our Local Authority and the school during this period. I finally had to get on my 'high horse' and remind them that they were in total breach of their legal duties.

I feel it's so important to tell this story as Harley couldn't fight for himself and he was unfairly discriminated against despite his diagnosis.

Fortunately, Harley's behaviour has really improved since becoming an adolescent. Like many teenage boys he's addicted to his X Box and YouTube. His literacy and numeracy is well behind for his age and homework is a massive struggle. As a parent of a child like Harley, I firmly believe that some children shouldn't have the pressure of homework.

My experience of being a parent of a child like Harley has made me absolutely certain that our education system needs a huge shake up for certain children with special needs. In my opinion there should be special centres for children like Harley - to do more hands-on skills and learn a trade. Harley is so very clever at absorbing things that interest him, and he could thrive in a less academic environment.

The lack of support and understanding I've received since Harley's diagnosis of 22q11 has knocked me for six. I have spent a lifetime fighting and it's been an incredibly lonely and isolating journey. I've also had to encounter prejudice from other mothers, neighbours and teachers. At times it's made me doubt my parenting. As to what lies ahead; not a day goes by where I don't worry Harley's future; particularly around his employment opportunities and most importantly his emotional well-being.

# 22q11 syndrome All-party Parliamentary Group

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