

The History of the
WILLIAMS SYNDROME FOUNDATION

williams
SYNDROME
FOUNDATION

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Published: November 2012



by Cynthia Cooper

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by *Cynthia Cooper*

In 1976 our daughter Clare was 14. Although she had no problems after birth in 1961 (three weeks late) at six months she started vomiting a lot after meals and became very dehydrated. Eventually at eight months a consultant announced that she had Infantile Hypercalcaemia (IHC) which might lead to problems with development. Naturally we knew nothing of this condition which entailed feeding her a low calcium diet to reduce the high calcium level in her body.

We discovered "Locasol", a tinned milk powder produced by Cow & Gate which had to be made up with distilled water, only available from a medical centre. We were also given a list of calcium-free baby foods which were quite restricting.

Within a few months her calcium level had returned to normal and fortunately never rose again, though she became very difficult to feed in the early years. A long procession of check-ups, assessments and educational psychologists followed over the ensuing years, with a variety of schools, but not too many medical problems.

One day in 1976 my neighbour, who was our local optician showed me an entry in a medical journal he had spotted, giving a brief description of hypercalcaemia. Never had doctors given us any information on the subject, nor had we ever met another child the same as Clare, since doctors seemed reluctant to put families in touch, even if they had come across another family which was unlikely with such a rare condition (now quoted as 1 in 25,000).

With hindsight I realise it was because they genuinely did not know anything about Infantile Hypercalcaemia since it had only been classified as a syndrome (under the name of Williams Syndrome) in 1961 – the year of Clare's birth.

I decided it was time I tried to find out what, if anything, had been written about it and was considering this when a friend called round. I told her what I was thinking of doing and said jokingly "perhaps this will be my life's work". And I suppose in a way it was rather prophetic.

In that year my husband George was working in London, on military affairs, so we had a temporary flat in town. I went up for two nights most weeks to keep an eye on him and make sure he was fed and watered!

This gave me the time and opportunity to do my research. I started with the Marylebone Medical Library in the Marylebone Road and looked up every reference I could find to Infantile Hypercalcaemia. There were not many, but I noted down the names of any doctors on the relevant medical papers which gave very little information and took my list back to the flat.

The doctors concerned were all at different hospitals, but I sat beside the telephone and tried to contact them. I managed to speak to quite a few of and after explaining my situation I asked if they might be interested in instigating some research into Infantile Hypercalcaemia. I suppose they must have been rather surprised – but maybe intrigued by the idea – so I crossed London in different directions to meet those who had expressed an interest.



Professor Tom Oppé

COURTESY THE TIMES 17 AUGUST 2007

By that time I knew that there were other families around for the doctors to study as we had already located 50. In 1976 my first effort had been to put a small ad in the *Daily Telegraph* asking for any family with a child with Infantile Hypercalcaemia to make contact.

To my delight I had five replies and shortly afterwards managed to meet two of the families here at home – both with children slightly younger than Clare. We were all astonished to see how similar the children looked. Clare had seen a photo on my desk of one of the children before they arrived and said "who is that little girl that looks just like me?" it certainly was striking and something none of us had realised before. There were of course so many other similarities in medical history and personality etc. which we were all fascinated to discover.

I devised a leaflet with photos of four different children of different ages and a description of the behaviour and characteristics then sent them around the country to paediatric departments and special schools etc. This produced quite a good response and gradually we heard of more families until by 1979 I felt we had enough to try and have a meeting in London.

I said if I could get together enough IHC children for them to study could they form a committee of other interested doctors and set up some research. I thought every angle of the syndrome should be covered, so I asked a paediatrician at the London Hospital, Dr Graeme Snodgrass; a cardiologist Dr Hallidie-Smith at the Hammersmith; a metabolic expert, Professor Bob Cohen; a psychologist Dr Bill Yule at the Maudsley and Professor Tom Oppé at St Marys, Paddington, who had written in the early 1950s papers on the rise in hypercalcaemia in babies at that time, due to extra vitamin D being added to milk powder. Professor Oppé, being the most senior, offered to chair the medical panel thus formed and George and I attended a historic first meeting at St Marys in 1980.

HAVE YOU SEEN A CHILD LIKE THIS?



8 months



6 years



7 years



9 years

The first family meeting was held in 1979



We were able to use Clare's school near Victoria (for £5 for the day) and 17 families came with children aged from 23 months to 28 years. I rallied my friends and relations to take charge of the children in the classrooms and persuaded three doctors to come and see them and realise what some of the problems were. Then one doctor gave a talk to the parents although at that stage not much was known. That doctor was Doctor Kershaw who was briefly on the medical panel but later dropped out.

We had another family meeting the following year and by 1980 fifty IHC cases were known. Our parents meetings had been attended by a medical journalist Pauline Walton who wrote for the "Doctor" newspaper given free to surgeries and after the third meeting she published an article with a photograph of all the children highlighting their amazing similarities.

This was seen by a freelance journalist called Brenda Marsh who contacted me with a view to writing an article on the whole subject which she hoped to publish in the national press.

By 1979 I was sending out Newsletters to all the parents who had contacted me and in 1980 we started an annual sub (£1 to start with) to help towards costs.

I designed a logo for our letterheads, the first of four over the years. It was used on all literature, magazines, lapel stickers etc. The design was intended to represent a child knocking to be let in (to the Foundation) and incorporating the HC of Hypercalcaemia. When we realised the connection to Williams Syndrome, we then added the WS before the original one.

All were in our "signature" colour of royal blue which we also used for the covers of our A5 size magazines from 1984 to 1996.



In 1996 the logo was changed again and in 1998 to the present one. Let us hope this final one is now here to stay.

While we were using the second of these logos a kind uncle of mine who did

some printing at his home offered to produce postcards and compliment slips with our logo in blue. This he did for me, entirely free, for many years and posted off batches to me when required and they were extremely useful.

In 1980 we had officially formed the Infantile Hypercalcaemia Foundation and become a registered charity with a committee of interested parents chaired by George and with me in charge of the Parents Association ("By Parents for Parents"). So we were a bona-fide organisation and could apply to charitable trusts for grants. The Duke of Norfolk had kindly agreed to be our Patron. We had ventured to ask him as he had been George's boss for a year in army days.

Brenda Marsh contacted a brilliant photographer, Terry Fincher, who she knew from previous assignments and he agreed to help.

I rallied several families to come to London and Terry took a series of photographs of the children. The combination of his wonderful photos and Brenda's article achieved miracles and in August 1981 there it was, the whole centre spread in the *Daily Mail*.

It was the best publicity we could have hoped for and resulted in me doing several radio broadcasts and a film on *TVAM* and *Newsnight* and an American TV show called *Monitor*. Rebecca Sobell flew over from USA in September to film it. Without this early publicity it is doubtful whether the Foundation would have grown and expanded at the rate it did.

The *Daily Mail* article caught the eye in other countries where it was reproduced, in particular in a free newspaper distributed to American supermarkets. A deluge of letters from USA followed which took up all my time to answer.

In due course an American Organisation was formed and we sent them a loan of £250 to help get them started.



The image that appeared in the Daily Mail, 1981

PHOTOGRAPH BY TERRY FINCHER © PHOTOGRAPHERS INTERNATIONAL

A local mother of an IHC baby girl came along to help with my rudimentary filing system and I managed to keep up with the correspondence. I wrote an A5 size leaflet describing the looks and behaviour of IHC children and attached a mini-print in colour of a little boy called Paul who had the typical "look" and beguiling smile. Paul had died sadly

at the age of 5, but I was so taken by the photo that I asked his mother in New Zealand if we could use it for our very first publicity leaflet. She very kindly agreed so our first "Paul" leaflets went out to every person and organisation that we could think of to appeal for money for research. I attribute a lot of our early donations to that appealing little face of Paul.

Joy Cooper wrote at the time as follows:

We are deeply moved that you should wish to use Paul's photo to aid the campaign. Your descriptions of the conditions are so alike that you might almost have had Paul beside you as you were writing. As I mentioned to Cleone after Paul went, I would be interested in the progress of your group. Thank you very much for giving us the opportunity of being involved in this small way. I know Paul would have been delighted – to him everyone was his friend.

Paul was a very special little boy who has left us with so many happy memories, and even now after this time has passed, little incidents occur which bring him close again.

Paul never really asked for anything for himself except to be loved. I would like to wish you and this venture well and feel certain that it must be successful. Best Wishes, Joy Cooper

I replied to Joy Cooper:

Dear Mrs Cooper, I was very touched to get your nice letter and quite overwhelmed that you should feel the way you do about the leaflet I wrote about your little Paul. He has such a cheerful and appealing little face and apart from being quite irresistible I feel that he personifies all that is best about our children. Somehow their charm and compassion and affection are all summed up in that infectious smile and I feel there would be no one better than him to help make the world aware of his plight and the problems of all the other IHC children like him.

I am so grateful to you and your husband for letting Paul help in this way and for being so understanding about it all. You – and he – will have played an enormous part in our publicity campaign when we really start spreading the word around and trying to get people to understand the very special problems and needs of our children and contributing accordingly. Yours ever, Cynthia

In 1981 we had 90 families and our funds stood at £7,000. We started to form regional groups around the country and also sell Christmas cards, which John and Mary Dunne undertook to distribute. This eventually produced around £2,000 p.a. and continues to this day.

By this time I was sending out a questionnaire to every mother who contacted me, to fill in details of their child. This was to

try to establish if any common cause could be attributed to the incidence of the Syndrome, from background, geographical area, early history etc. It soon emerged that there were so many characteristics in common the most striking being the sensitivity to loud sudden noises (hyperacusis). Fireworks, balloons, guns etc were and still are a no-no for IHC children and 98% were eventually calculated as suffering from this problem, something never before discovered.



Paul

Meanwhile I had been contacted by some families in New Zealand who had read our publicity but their children, despite having the same profile, were diagnosed as having "Williams Syndrome". This was because a Doctor Williams in Auckland had written a paper describing the medical characteristics of a small number of children with aortic stenosis (a typical heart condition) and identical facial features.

It dawned on us that this was, of course, the same syndrome that we were dealing with and we had, by then, discovered that not all babies had an episode of hypercalcaemia in infancy, so decided to rename our Foundation to coincide with the rest of the world, in particular USA, the Williams Syndrome Foundation (WSF).

This is now recognised internationally. Thirty one countries now have their own WS organisation and information continues to be spread far and wide.

In 1982 we had our first Hyde Park picnic with 18 families which we videoed. The picnic, now in Regents Park, continues annually and is very popular.

When our medical panel had finally come together and discussed future possibilities,

Doctor Snodgrass introduced Doctor Neil Martin who was interested in starting research. From 1982 to 1984 Neil went around the country interviewing parents and amassing information at the end of which the first ever in-depth paper on WS was published. This research was funded by the WS Foundation.

After this, research was ongoing and the Foundation had to raise many thousands of pounds to fund all the projects. Action Research, a grant making trust, was our first major donor. Research still continues here and in other countries although the significant breakthrough occurred in 1993 with the discovery of a micro deletion of the elastin gene on chromosome 7, which appeared to be the cause of some, but not all, the characteristics of the syndrome. As a result of this for the first time ever it became possible to prove to almost 100% accuracy a diagnosis of Williams Syndrome through the FISH test, a simple blood test (Fluorescence in situ hybridization).

We were still sending out information to paediatric departments, including some large 3x4ft black and white posters of a collage of children's faces, since recognition is the key to diagnosis.

I also thought we should make a video to distribute information around the country. So with the help of Sam Holloway, a life-long friend, and his photographer colleague, we produced a 20-minute video of a selection of families and doctors to give a good idea of the syndrome. We then sent as many as possible to paediatric departments around the country in 1991.

Around this time several local TV stations interviewed families as information and interest in WS spread. A new video was produced by the Foundation in 2000 to add to our collection. These are all held now in our archives in DVD form.

In 1983 we had our first day meeting in Bedford college London which was well attended. Also in 1983 I thought it would be particularly helpful for families with young children to experience shared holidays. We went to visit Gorslywd Farm in Wales, a well run conversion of eight cottages varying in size, run by a sympathetic couple. It was near the sea, away from main roads and so peaceful and completely safe.

The Foundation subsidised the holiday weeks and they were a great success. All the cottages were booked for many years and a great many parents paid tribute to the benefit of being with other parents in the same position and of course the children loved having the company of others.

Sadly a few years ago Gorslywd ceased being a holiday centre, though recently it was discovered to have started up again under new management and this is currently being investigated. Suitable similar establishments are hard to find. Another system we set up which was even more beneficial was holiday weeks for adult WS people.

We found two locations offering supervised holidays, one in Cornwall and one in Norfolk and these have been enormously popular for the over 18 age group. They have been constantly fully booked since started in 1988 and there is great disappointment in this year of 2012 that the Norfolk home, Rainbow is having to close down at the end of the year for financial reasons. It is hoped to find an alternative but it will not be easy. Rainbow was immensely popular and will be very hard to replace.

In 1991 and 1992 we produced a booklet "Guidelines for Parents" (illustrated by me!) and the following year "Guidelines for Teachers" produced by Orlee Udwin and Prof Yule and in 1996 "Guidelines for Families and Professionals" by Dr Udwin, Dr Howlin and Mark Davies and also "Guidelines for Employers". All of these over the years have been extremely helpful

In 1988 George and I were invited with Clare by the US WS Organisation to attend their bi-annual Convention in Salt Lake City. We were given a wonderful long weekend and met their organisers and doctors and many families and it was most interesting to exchange information etc.

Two years later, in 1990, we undertook a major expedition to Australia and New Zealand. Meetings of their local families were organised for us in Sydney, Auckland and Wellington and it was fascinating to meet the people with whom we had so far only corresponded by letter.

One-day conferences at Bedford College became an annual event and were always fully attended. In 1989 George and I were overwhelmed to be presented with magnificent presents for our efforts in setting up the Foundation; beautiful watches each and a lovely silver coffee set. It certainly was an unexpected and most generous present from the Foundation.



Hyde Park, 1982

That year we also started giving an annual “outstanding” certificate to the aims of the “WS IHC Foundation”. The first recipient was Pauline Barnett who had tirelessly been raising money from her garage sales and up to then had raised £6,000. Her final tally before retiring was £40,000.

By then also the three holiday week locations were continuing: Barrows Green in Kendal for children, Gorslywd in Wales for families plus Rainbow in Norfolk and Trevanion in Cornwall for unaccompanied adults – still as popular as ever. In 1991 our annual picnic changed to Regents Park which was equally popular.

In 1992 Mr Pendle, managing director of Cow & Gate decided to resign.

Earlier they had made a proposal to alter the formula of Locasol, which would entail mixing six ingredients instead of just milk powder and distilled water. We rallied our doctors and the dietician from Great Ormond Street and argued that parents of WS babies had enough struggle already with feeding difficulties without these extra problems. Mercifully our arguments prevailed and Cow & Gate withdrew the suggestion and even agreed to incorporate contact information for the Foundation on the Locasol tin labels.

In 1993 Mr Pendle was entertained to lunch by the committee and he responded with a donation of £2,000 to the funds. We parted on excellent terms and wished him well as he set forth for pastures new.

Also in 1992 we arranged our first ever weekend Convention in Stoke on Trent in the Stakis Grand Hotel. It was fully booked and all sessions were well attended. All our professional panel of doctors came and also a doctor Bdzuch from Czechoslovakia with whom I had corresponded. We had invited the Lenhoff family from the USA as their WS daughter Gloria was a wonderful singer. She sang in many languages and was renowned in

her country. At our gala dinner she sang wonderfully for us and our British contribution was Jeremy Colborne on his keyboard and Andrew Skinner playing his portable organ.

Altogether it was a wonderful evening. The whole weekend took a huge amount of organising. Jill Robinson and I strained our brains working out all the details but in the end it was voted a success. All the Williams children and adults were taken on outings all day and the babies cared for in a crèche in the hotel. Luckily nobody was lost or left behind and they all survived to tell the tale.

Back in 1988 Doctor Hallidie-Smith discovered a vascular abnormality in the collagen of WS children and this was the first sign of the eventual discovery in 1993 of the micro deletion of the elastin gene on chromosome 7. By this time Doctor Dian Donnai who had been doing invaluable research in Manchester had been working on this for us and so the FISH test was produced and could prove if the deletion was present in WS people. This is now the standard test for diagnosis of proof of WS throughout the world. It was a most significant breakthrough.

The deletion does not account for all the effects of WS and research still continues in this area. In 1995 sadly Doctor Hallidie Smith died but she had been able to see the results of her earlier findings. She still is most sadly missed. She was dedicated to her job and never refused to see a WS adult, even though she was a paediatric cardiologist.

In 1993 we had the great good fortune to “discover” Doreen Goodall who lives locally and agreed to become our “Administrator”. This meant putting all information onto a computer and generally bringing the Foundation into the 21st century (slightly in advance). I still maintained I could put my

finger on someones name/address/date of birth etc. on my card index system faster than her! I won many a race against the computer. But she became an enormous help and gradually took over the office and came to know many WS families.

She took a real interest in all our activities and attended meetings, conventions and the annual picnic. After a bit I wondered how I had ever managed without her and she and her husband continue to be good friends. Doreen finally retired from the WS in 1998.

On the appointment of John Nelson to Chief Executive, an office in Tonbridge was acquired and manned by him and his wife Pam. Pam’s job was eventually taken over by Sarah Green and John handed over to Lizzie Hurst.

In 1993 Tom Oppé had decided to retire from the Professional Advisory Panel after 12 years as soon as a successor was found. This fortunately Neil Martin undertook to do. At the Regents College meeting that year George presented Professor Oppé with a porcelain owl (for wisdom) from the WSF.

George announced that year that he and I would be retiring in 1995 and we were very lucky that Mike Adlam had offered to take over as Chairman whilst Doreen continued to manage the Parents Association. So we stood down and passed the baton, knowing the WSF would continue in good hands.

By 1999 a considerable number of European countries had formed their own WS organisations and Susie Cooper initiated a joint membership with the UK to facilitate the sharing of information. This was entitled Federation of European Williams Syndrome (FEWS). The initiative for this came originally from the Italian AISW and was first mentioned at an international Convention in Rome by Leopoldo Torlonia, father of a WS daughter.

In 2004 formal acceptance by the EU was obtained and FEWS was officially recognised as a registered charity. Susie

Cooper is the current chairman with Anne Breen (Ireland) the treasurer and Paul Pyck (Belgium) as secretary. In 2012 a sponsored bike ride to Paris raised around £12,000 towards costs. Annual FEWS meetings have been held in a variety of countries since, with fully paid up membership currently standing at 15. Summer camps for WS children are also held in varying countries with financial help from the EU. These have proved very popular and Guy Morgan and Freddie Wolfman have each been to one.

The 2012 summer camp was held in Spain and the annual meeting in Leuven, Belgium. Also in 2012 a calendar was produced for distribution in Europe to raise awareness, and these will be repeated annually and distributed internationally.

Back home there have been more conventions. In 2000 the bi-annual WS magazine doubled in size and is now a very smart and professional A4 size production, the cover being a collage of coloured photographs of families and events around the country over the preceding year.

In July 2004 a Silver Jubilee Convention was held in the Park Hall Hotel, Preston. George and I attended and it was great to see so many old friends again from our earlier days.

Mike Adlam has organised and performed in many gruelling fundraising marathons and events over the years which have added enormously to the funds and parents continue to do sterling work keeping the coffers filled.

There have been many changes of committee members and doctors since we left and became joint patrons instead of the Duke of Norfolk who sadly died in 2002. Neil Martin continues to head the PAP which considers all research projects to be submitted and Mike Adlam still holds the reins as Chairman as at the date of writing.

Cynthia Cooper