MY PERSONAL EDS HISTORY

(Not to be considered as the ravings of a hypochondriac but rather to be an aid to the understanding of the manner in which EDS has affected the lives of some of those marked in the family tree as direct descendants of the Fletchers of Ightenhill.)

Looking back I can realise that Ehlers-Danlos Syndrome changed the course of my life from the moment I slid down the side of an old fashioned haystack and dislocated my left knee cap. An ambulance was called for me and the dislocation was reduced. From that day on my life was changed, I was different from my peers and often in pain.

Following that first dislocation I was away from school for many months and no longer allowed to take part in any physical activities or games. When my friends were going to dances, playing tennis or taking up sports I was unable to join them and this excluded me from many social activities they enjoyed.

Once I had left school I found that my chosen special interest of woven textiles was made difficult by my weak muscles and tendency to muscle spasm when operating the pedals of the looms. I found ways of overcoming this but it remained a problem right through my student days. It was about that time that I first started getting tenosynovitis affecting my hands and arms. Additionally I found my shoulders painful if I lifted my arms above my head. It didn't occur to me that the problems with my hands,shoulders and knees could be connected.

Since there are so many ways that the Ehlers-Danlos Syndrome (EDS) can affect the body, the clinical diagnosis is difficult and doctors are now urged to treat all EDS patients, regardless of type, with extreme care. The inability of the abnormal collagen fibrils to organize into bundles that are essential for the formation of a strong network results in defective type III collagen, a protein that is expressed in many tissues but is primarily a component of extensible connective tissues such as skin, gastrointestinal tract, bladder, uterus and the cellular structures of liver, lungs and the vascular system.

If EDS is difficult to diagnose it is even more difficult to type, I was in my 40s when I was first told that my unknown heritable disorder was named Ehlers Danlos Syndrome. I had complained about hand weakness and problems with grip and was told "You have to expect that because you have Ehlers-Danlos Syndrome".

No one had mentioned that name before and when I finally got the diagnosis confirmed at age 62 I was told that I have the Hypermobile type. Once the diagnostic criteria were changed my diagnosis was amended to Classical type (because of skin involvement and papyraceous and wide scars) but with elements of the Hypermobile type.

Recently a respected researcher in EDS has said that Classical and Hypermobile types blend into each other and it makes no real difference what it is called. Hospital doctors are advised against assuming that a patient has been typed correctly and to treat all EDS patients with extreme care. A positive family history and symptoms matching those in the 1997 criteria for the recognition of EDS are sufficient cause to seek further advice.

One thing is certain and that is that the form of EDS present in a family will stay the same even if the degree and the manner in which individuals are affected is very different. It is the extreme variability in the severity of symptoms in different individuals with the same type of EDS and the varied manner in which the weak collagen causes the symptoms that explains why the EDS appeared to skip my mother's generation and to have been present only very mildly in my grandmother despite the fact that it is inherited as a straightforward (not sex-linked) dominant in our family.

With hindsight I can see that EDS manifested itself in both of them-my grandmother had a spinal curvature probably due to muscle laxity, loss of bone density and severe arthritis. She also had the skin typical of EDS, lax joints, a heart problem never fully investigated but maybe a valve problem common with EDS.. She also had recurrent urinary tract infections and chronic bronchitis. She suffered a number of fractures, one of them resulting in a contracture of a finger. She had two pregnancies but the second resulted in the premature birth of a baby boy who died at about nine days old.

My mother had undiagnosed chest pains, symptoms of hiatus hernia, early onset arthritis and lax ankle joints. She suffered severe gum disease following Scarlet Fever as a young adult resulting in the loss of a

large number of teeth. After her first pregnancy she developed varicose veins and the discolouration of her lower legs so typical of EDS was diagnosed for her as “varicose eczema”! My sister and I both developed the same discolouration. In her case it has been attributed to varicose veins but in mine it is the result of many large haemotomas. Photographs taken of my mother in her 20s show her to have the Marfanoid habitus and to be standing in typical EDS stance.

In later life she suffered badly from unexplained dizziness and malaise and her back became so stooped that if seen from behind her head was not visible at all. Now I am in my mid seventies and going the same way I can understand that she probably had the same difficulties with osteo-arthritis, osteoporosis, weak muscles and pain as I have.

Both my mother and grandmother complained of fatigue but this was never taken seriously by their doctors or family. It is only recently that chronic fatigue, bone loss, autonomic dysfunction and all the other problems they suffered have been understood to be features of EDS. There was no disabling hypermobility disorder in either of them although my grandmother explained that she and I were double jointed and that it was a family characteristic inherited from her father. She was proud of it and, for a while, so was I.

My grandmother had three sisters and three brothers. One of her sisters passed the condition on, another does not appear to have done so(although my grandmother told me that all her sisters were double-jointed.) and the third was almost certainly affected but was childless. Two of her brothers died without having children and the third, the eldest may have passed the condition to his son who died of a heart problem aged 21 and may have had EDS and been the “crippled cousin” my mother told me she used to play with.

Joint laxity is present in several descendants of the eldest sister and from records and family recollections it is clear that the EDS was present in my family for many decades but that not all of those with the genotype had disabling hypermobility.

I have had contact with “double-jointed” distant cousins whose shared ancestors are of the same as mine and who have varying degrees of joint laxity themselves or know of family members affected. Joint laxity has proved to be the best marker to study as few family historians would share details of their medical problems.

EDS first affected me as a very young child when I was slow to stand and walk and was described as having “outgrown” my strength” because I was tall for my age and thin with a pale complexion and soft easily bruised skin. My skin was once described by a doctor as like “uncooked pastry” After a warm bath my blood “pooled” in my feet leaving them a dull red. I was able to squat on the floor and twist my legs and feet sideways and I could twist my arms right round my back. My elbows bent the wrong way and I was told that I would make a good gymnast. Even in childhood I became easily fatigued and fainted on several occasions when required to stand for an unusually long period.

At the time I was prescribed iron tonics and my parents were told that my entry to school should be delayed by one year. I had the typical pain of EDS hypermobility which was diagnosed as “growing pains” although it continued through adulthood, long after I had stopped growing. In fact I grew very tall, with extremely long arms and legs. These were not only been a problem to me when shopping for clothes but are now considered by some researchers to be a feature in EDS hypermobility- the “Marfanoid habitus” with a “wingspan” greater than overall height. I also had a nose with a bump where bone and cartilage met. Never would I have believed that this was due to EDS but in 2004 it was described as such and noted that many woman patients have already had surgery to reduce such features. That is exactly what I did. The same research paper mentioned the frequent occurrence of deviated septum in connective tissue disorders. This was another of my problems eventually corrected by surgery.

I was eleven when the first dislocation occurred. From that time on dislocations of both patellae (kneecaps) became common and could be caused by simple movements such as bending to stroke a cat while having one hand on the knee. Despite the fact that most literature on EDS suggests that the dislocations can be easily reduced mine were so complete that reduction under anaesthesia was required each time. Each dislocation was followed by weeks of inactivity as the ligaments were torn and the pain and swelling persisted. ! was told to wear supportive crepe bandages on both knees all the time and these were a source of great embarrassment as they showed beneath the hem of my school dresses.

I was given a wheelchair and kept off school for several weeks each time I had a dislocation. Eventually I had surgery to transfer the tibial tendons further down and across the tibia (shin bone) on both legs. This was when I was seventeen and I was in hospital for several months. I had to miss college for a year while

learning to walk again. At the hospital the surgeon talked to my mother -not to me! .She later described it to me as a disease where “the muscles at your front are too loose and those at your back are too tight”.

Unfortunately the transplanted tendons stretched after a time and dislocations resumed. They were less severe than before so that I could reduce them myself. Following these later dislocations there would be pain and swelling causing me to use walking sticks to move about. Eventually the screws used to secure the tendons in their new position caused inflammation and were removed.

When I was still very young I was advised that my internal organs would be more fragile than normal and advised to avoid pregnancy because of risks of uterine rupture and of passing the disorder to future generations. I was told to expect my muscles to be weaker than normal and that my bones would be more susceptible to fracture. No one named my disorder as Ehlers-Danlos Syndrome although I now knew that it was inherited. I told my mother that I felt that was from her side of the family and she was so devastated that I never mentioned that aspect of the problem again.

Spontaneous effusions in the knee joints became frequent and necessitated trips to hospital and almost permanent use of walking sticks. A lumbar scoliosis had been noticed when I was 18 but physiotherapy gave no improvement.

From about the age 25 I experienced extreme fatigue even after stopping activities that I thought were the cause. In the 1980s this was diagnosed as Chronic Fatigue Syndrome. Only much later did I find that chronic fatigue is a condition caused by EDS.

By my late 20s I had recurring pain in my chest and upper back subsequently diagnosed as costochondritis (a feature of EDS) and vertebral instability. A disfiguring curve in my upper back was apparent and at the outermost point the rib would dislocate. I used to visit a chiropractor to help with this until I was told that it was too risky for him to treat me any more.

Numerous fractures occurred and osteo arthritis started to develop in my early twenties. Probably because my hands and wrists were over stressed by use of walking sticks and from pushing up my body from a seated position I gradually developed severe problems with my hands and wrists. Investigation showed that as well as recurrent tenosynovitis there was nerve compression, soft tissue rheumatism and a degree of osteo arthritis. Although I have had surgery several times these problems are still present and gradually getting worse.

By the time I was 30 arthritis had become became so severe in my knees that removal of the knee caps was advised .This was the only treatment available at that time. I was told that my patellae were so damaged that they had been photographed and the one from the left leg had been added to the collection of the Royal National Orthopaedic Hospital museum. Recovery was not complete due to the muscle weakness of EDS and I needed a further tendon transfer to lessen the instability of the left knee joint.

When I was 40 I was officially registered as disabled and a wheelchair and a left leg calipers was provided. Unfortunately this set up problems described as sacro-iliac dysfunction and I stopped wearing it. My consultant recommended isometric exercises. (now strictly contra-indicated). Eventually I was able to walk with sticks again and the absent knee caps could no longer dislocate but the ligaments and tendons were still loose causing my legs to curve backwards and to be very unstable so that without warning, the joints would apparently twist out of position causing effusions and long lasting pain..

The recurrent spontaneous effusions in both knee joints continued for twenty three years during which time I was also diagnosed with various problems of the stomach, bowel and urinary system and developed a hiatus hernia. I had an episode of severe chest pain first diagnosed as pleurisy but eventually diagnosed as spontaneous non-tension pneumothorax. This latter condition is a symptom of EDS although I didn’t know that at the time.. My grandmother had been diagnosed with pleurisy on more than one occasion and I wonder now whether she had the same problem.

It was found that I also had skin, inhaled and ingested allergies and I was put on permanent medication for “migraine.” Changes in the bony structure of the hands and wrists became worse and the osteo arthritis, tendonitis, and bursitis caused pain and loss of movement in the hands, arms, shoulders and hips. I had regular injections of steroids to allow me to continue normal life and my hypermobile spine required me to seek regular treatment. Each time I had surgery there was a post operative bleed and in most cases the wound healed slowly with the tendency to gape open despite careful stitching or stapling.

There were also numerous minor fractures in the toes, feet, ankle, and wrist until in 1987 I fell and broke the neck of my left femur. Because I was deemed too young for a “hip replacement” the bone was repaired with a pin. Unfortunately the pin punctured a blood vessel resulting in avascular necrosis. Orthopaedic surgeons are warned to take great care with EDS patients because the bone density is reduced but no one concerned knew that at the time. As a result I was wheelchair bound from 1987 until 1994 when I had a total hip replacement followed eventually by two total knee replacements..

I had hoped that the three “new” joints would allow me to walk normally but the combination of weak muscles, poor balance and continued instability of my knees without their patellae prevented that and the use of crutches or walking sticks caused many problems with my defective shoulders, arms and hands.

About that time I began to experience severe pain in my lumbar spine and investigation revealed, facet joint disease and osteoporosis as well as the known scoliosis. Those problems when combined with the increasing inability of my muscles to hold my back upright despite physiotherapy to strengthen them and my unstable knee joints eventually led to full time wheelchair use again. I had also developed autonomic dysfunction causing my blood pressure to drop suddenly so that I could only stand still for a few minutes without feeling ill and faint.. This is now well known to be a problem in EDS and thought to result from weak walled blood vessels affecting circulation. In my case it is now treated successfully with steroids.

I now know that all the conditions I have mentioned are all thought to be the result of EDS but in my case many have been made worse by lack of medical knowledge about the ways EDS can affect the body and by what are now known to be the wrong treatments. Together they have had life changing effects. I was denied a normal social life as a young adult, I followed the advice not to have children, I lost my career and by the time I was 42 I was on a disability pension and under pension rules not allowed to work at all.

Fortunately there is now a great deal more knowledge of the heritable diseases of connective tissue and the life of a child born with EDS today can be very different to mine. However very few family doctors know much about EDS and it is best to provide them with background information to demonstrate that there are many more effects of the syndrome than stretchy skin and generalized joint looseness and also that the effects do not have to be severe for the syndrome to be present.

Most effects of EDS are also found in the general population and can be treated in the usual manner although surgery needs to be approached with caution. If a number of the conditions mentioned here or in the list of signs and symptoms given in the article below occur together AND there is a direct relationship to a family member diagnosed with EDS it is best to contact one of the EDS Support Groups. These are the Ehlers Danlos Support Group in the UK and the Ehlers Danlos National Foundation in the USA . These organizations offer valuable information on the syndrome and both of them have a bulletin board.

Pat Newton
26 May 2007.

ANOTHER PERSONAL EDS HISTORY

My problems first began with my throat, at the age of 13 my throat became very lax, which made it impossible for me to sing at school. I had to drop out of the school choir and as years have gone by this has continued to be a problem causing me to lose my voice frequently and I always have what I call “a sticky throat”. Anaesthetics have been a particular problem because of this.

.

In my early 20s my right knee began to subluxate and in my middle 20s I developed severe problems with my back. This was originally diagnosed as osteochondritis and I was prescribed spinal corsets which I wore for 28 years.

When MRI scans became available I was sent to London for this to be done. At that time the consultant told me he could help to a degree and I was in full body plaster cast for 12 weeks followed by lumbar fusion surgery. This gave some

improvement but it was short lived and to this day I am still affected with muscles being so weak that just “hoovering “or leaning over the sink causes a great deal of discomfort and causes my spinal muscles to go into spasm.. (It was while I was recovering from the spinal surgery that in discussion with my sister over the similarity between some of my symptoms and hers that we realised that I might have Ehlers Danlos Syndrome myself. This was later confirmed when I was 56.)

At the time I was registered with a GP in Lewes named Dr Rowland and he demonstrated how I could help myself by lying flat on the floor(nowadays it has to be a table) holding my arms up, together ,and slowly from above my head move them down to lie flat on my body. This I have to do several times a day whenever I do something, even if it’s only standing for a while or walking.

At the age of 40 my mother was diagnosed with Osteoarthritis in both knees although she was told that it was simply “the bones wearing out” and made a joke of it until it became extremely painful. When I was 40 I too was diagnosed with osteo-arthritis of the knees and have since had a total knee replacement to my left knee and patella femoral surgery to the right.
.
I have had 9 fractures ranging from fingers wrists and elbows ,which before they were broken would subluxate, I dislocated my right shoulder and have rotating cuff injuries to both left and right., and have steroid injections every 6 months ,which does give some relief

Varicose veins started in my 20s ,I have had them injected ,cut and tied ,and finally stripped, on 2 occasions I have had a vein burst ,I am now wearing elastic stockings .My lower legs are discoloured in exactly the same way as my mothers were so many years ago.

Raynauds Syndrome has been another problem in my life and for that I have had bilateral sympathectomy surgery. Like my sister I have had bilateral carpal tunnel release surgery and my circulation has been described as vaso spastic.

Healing after operations is always a problem for me with stitches having to remain longer than normal and teeth extractions have to be done at hospital due to excessive bleeding Additionally I now have a prolapsed bladder, which Dr’s don’t want to attempt to repair. At age 71 I have a great deal of arthritis pain in my right knee and left hip.

Rosalie Kay
7 August 2007

The Ehlers Danlos Support Group
http://www.ehlers-danlos.org/
The Ehlers Danlos National Foundation
http://www.ednf.org/