

Good [Greetings] Ladies and Gentlemen, today I have been given the honour and pleasure to be able to come and talk to you about my condition and my experiences with the health professional service and also about my transition from young person services to adult services.

First I will give an overview of the condition which I have. I have a condition which is known as Osteogenesis Imperfecta, (Osteogenesis – Formation of Bones, hence Osteogenesis Imperfecta in literal terms is the imperfect formation of bones), it is more commonly known as Brittle Bones Disease. OI is a condition which causes extremely fragile bones. OI is a congenital disease; this means that it is present at birth. It is frequently caused by a defect in the gene which produces type 1 collagen, collagen is a group of naturally occurring protein, this is essential for strong bones and muscle structure. There are many different defects which can affect this gene. The severity of OI depends on the specific gene defect. This is why there are 8 different types of OI ranging from Type 1 to Type 8. I have Type 4 OI. On the presentation behind me you can see what other symptoms there are for the other types of OI.

In most cases OI is inherited from the parent as it is a dominant disease but in some cases is the result of a new genetic mutation which was what happened with me, no one else in my family has ever contracted OI so for my family it was a whole new experience, I shall talk more about that in a few minutes. A person with OI such as myself has a 50% chance of passing on the gene and the disease to their offspring.

People with OI are most susceptible to fractures than normal people and usually they are below average height but luckily for me that is something which hasn't affected me. I am a normal high of about 5 ft 6 inches. The most common symptoms of people with OI are they have Blue tints to the whites of their eyes (Blue Sclera), Multiple Bone fractures and they can experience a loss of hearing and typically deafness at a very young age, this loss of hearing happens normally between teenage years to being a young adult. I have to get tested for my hearing every year, I had my hearing tested last September and I am happy to report that my hearing is fine which always good news is. The loss of hearing can occur through fractures of the tiny bones in the ears. Also it is found with people with OI that they have poor development of teeth again I have been fortunate to not have that either. There are a few more noticeable symptoms of OI. Including bowed arms and legs as well as Scoliosis, which is basically the curvature of the spine in an S shape. Another complication which I have experienced which has been put down to my OI is respiratory problems, as generally most people with OI are confined to wheelchairs it makes it very difficult of them to cough and to clear their lungs as well as a person who can walk and is able to do that regularly. This has meant that I along with many other people have an increased risk of getting flu's, chest infections and even pneumonias. A couple years ago I had a bad time of it getting an awful chest infection which meant I had to be in hospital for over a week , on oxygen, IV antibiotics and IV fluids for more than 3 days it was not a nice experience, this means I have to be especially careful during winter times not only due to my bones being weak but also to avoid cold getting into my chest, to monitor the condition of my lung I have to go and get a lung function carried out every year in order to make sure they are functioning as they should and also I get chest physiotherapy in order to make sure I am as safe as possible during cold winter times when I am more likely to be at risk.

OI is obviously suspected in children whose bones break with very little force and also due to the blue sclera which I mentioned before but a definite diagnosis can be given through carrying out a skin punch biopsy and also family members may also be asked to provide a DNA blood test. This is normal in cases where there has not been a family history of OI previously. On the other hand if there has been a previous case of OI in the family than normally a chorionic villus sampling is carried out during the pregnancy to determine if the unborn child has the condition. Due to the variation of mutations which can occur to cause OI, some forms cannot be diagnosed with a genetic test. Some more severe cases of OI can be detected on ultrasounds when the foetus is as young as 16 weeks.

STATISTICS SLIDES * 2

Unfortunately however no cure for OI has been found, although there are drugs and other therapies which have been proven to help people with OI to reduce the risk of fracture and reduce the amount of bone pain. Luckily for me I do not experience bone pain on a regular basis thus is the type of my condition. Currently I am undergoing a treatment of Biphosphonates Pamidronate, which is typically used to treat people with Osteoporosis but this has shown very good improvements with people with OI, especially in children. This drug has been shown to increase the strength and the bone density of people with OI. I started receiving this treatment in November 2005 and since then I have experienced a great improvement to the quality of my life. Before I started this treatment I used to be in the hospital all the time, I would be in the hospital for 6 months at a time, the time in which nurses and other health professional became a huge part of my life. Before starting the treatment I think I could safely say I have over 100 fractures in my life time. Since beginning the treatment that number has greatly been reduced. For example an incident which happened while I was at Sixth Form College last year when I got knocked over in this chair by a girl running into me causing the chair to tip on its side with me still in it landing on my ribs before the treatment would have caused me to have multiple rib fractures and possibly a broken arm to boot but thankfully I managed to get away with a few bruised muscles and a slightly cracked rib so I was very lucky and fortunate there. Also for people with OI it has been shown that low impact exercises such as swimming can be used to keep muscles strong and maintain strong bones. Although in severe cases surgery may have to be implemented into the long bones of the legs in order to strengthen the bones and reduce the risk of fracture. I have had this procedure carried out on me when I was 2 years old. I have a metal rod from the top of my right hip bone down to my knee. Even though there are no permanent cures to OI I am happy to be on the medication which I am on now, I have to go in to hospital every 3 months and stay in for 2 days while I get the necessary doses of the medication. This may seem a bit tiresome but I suppose for me it is better than spending most of year in hospital.

Now I have gone through a brief summary of Osteogenesis Imperfecta I would like to tell you all a quick story about my life. When I was born I wasn't showing any symptoms of OI, I was a normal happy child, the only concern my parents had about me was that I wasn't able to walk but when they took me to the doctors they said that it was nothing to worry about and it will happen eventually about 8 month after I was born though I started fracturing my bones, multiple time one after the other. Back then OI was very uncommon condition and not many people had heard of it so one day when my parents took me in with

an injury, they got arrested as they believed that I was being subject to child cruelty and the social services wanted to take me away from my parent but my loving parents would not give me up, they said they would have to die before they took me away from them, which makes me tear up to this day. There was a tremendous court case which went on for a year or so before one day in 1996 2 years after I was born when people at GOSH noticed that something was not quite right when I was crawling along and I just fell in front of one of the doctors and they witnessed that my leg was swelling up and then they did the test which I mentioned earlier and discovered that I had OI. Which meant that my parents were freed but to this day the case has not been closed as the judge at the time said that there was not enough evidence. The reason I tell you this is that I want as many people to be aware about my condition and about OI so that no one has to go through what my parents went through and that's why I want to do as many talks like this as possible to promote OI and to raise awareness about it.

Now I will move onto talk about my experience with Health care professional, as you have seen so far I have had many experiences with professional in my past, be they OT, Physiotherapist, Nurses, Doctors. I feel that they have been a huge part of my life. Without their help I would not be able to be here right now as the man I am talking to all of you about it. They have helped me in every aspect of my life. In my short 18 years of life I have had about 20 physiotherapist, 25 occupational therapists and 6 different doctors look after my care. And for someone like myself who was unable to speak English till I was about 5 was quite a scary experience but I suppose what helped me through it and made me less afraid was how nice those people were and still are. That is the main thing whatever job you go into within the Health Sector being nice and friendly is the most important thing, this may seem like an obvious thing but when you are scared and in pain the most beneficial thing to a young person is a nice friendly face who they know that they can trust and is not trying to hurt you and is only trying to do the best for you. I can very confidently say all the doctors, nurses, OT and Physio's I have worked with are the loveliest of people to work with and they are very helpful. I can recall a lovely moment back in 2004 when I turned 10 I was in hospital unfortunately with a broken leg on traction but all the nurses who knew me decided they would throw me a little party thing in the ward which was such a lovely surprise one which I wasn't expecting. My parents bought chocolate and cake for the whole ward and we had a great time and it's something which I remember to this day. This goes to show those little lovely gestures which go on to make a big impression in someone life. It showed me that other people do care and people that I am not related to or friends with are really nice and are lovely people. I do though think of some of the nurses who work at Lewisham Hospital as my friends as I have grown up with them and I have seen them work up the ranks of being a new trainee nurse to becoming a head nurse on duty on the wards. To this day I remember every nurse and doctor who has worked with me.

Now transition, this is a big part of my life which is taking place right now. I have recently turned 19 in January which has meant that all the health professional services are moving me on from the younger adult services onto the adult services. Most of the transitions have taken place, the only one which remains is for the MAPP which is in charge of coordinating all of the services for me while I still a young person to hand over to the adults team, this will be happening within the next few months. This is why I was asked to come and talk to you about. Transition is something I was not looking forward to from the

very first time I was told about it when I was 16. As I mentioned before even though I have gone through many OT and Physio and other health care professionals in my life time due to the amount of work which they do for me in order to make my life easier and better has meant that I build up a very solid relationship up with them and it is very difficult for me to let them go. I suppose as you get older you need to learn to move on but due to the nature of their work with me and the amount of time they spend with me it is quite difficult. I shall give you an example up until I was 17 I was under the care of one Dr John Stroobant who was a child paediatrician who was in charge of my care for many years. He has been the doctor who has overseen all parts of my care from a very young age and helped my parents after the ordeal which they went through which I mentioned earlier and he a man who would come and see me pretty much every time I was admitted onto the wards at Lewisham to ensure I was getting on alright and to make sure everything was okay and I was getting on well with school and everything, he even offered to pay for my transport one year to go and get my exam results. When we were talking about transition and me being moved on to his colleague who is a rheumatologist who now takes care of my care I was very reluctant as Dr Stroobant has been with me through tough times and I didn't want to leave his care but I knew I must. I have to say though the new doctor I am under now is really good but that period of transition from him to my new doctor was a tough time. After that I realised I must be moving on to the other adult services from other teams as well, I suppose having gone through the process one it has sort of made it easier for me to know what to expect. Luckily thanks to the transition team they made the transition from each service very smooth. Also what helped me most was people who I worked with when I was really young as part of the children services were now working as part of the transition teams for the respective services which made the transition process that much easier, having familiar faces certainly helped.

Now looking to the future, the adult services are the people who will be helping me through a large part of my adult life. Looking in the short term they will be the team who will be helping me with my university choices and making sure the university which I go to has suitable adaptations for my needs and to ensure that the accommodation I will be staying in will be appropriate and I will have all the equipment I will need and I have all the care which I need. As I know and realise I will require help and support throughout my life and the adult services team will help to make that easier by helping me to sort all of that out. In the long term they will help me with getting a job and helping ensure I have equal and fair rights and they will be supporting me through adaptations that I will need at the place where I will be working and also if I was to buy a house they would help me with getting funds and grants in order to make the adaptations I will need. Transition is a big part of my life and I am nearly on the way of completing that part and I look forward to what the future will bring.

Thank you all very much for being such an attentive audience and once again it has been my absolute pleasure to have been allowed to come and talk to you about this, now please I do encourage any questions you may have on anything you can think of regarding my condition, my experience with my condition, my work with health professional or anything else which I may have mentioned throughout this talk. Thank you once again for listening.

CVS

Chorionic villus sampling (CVS) involves taking a sample of cells from the tissue of the placenta of a pregnant woman. These cells are called chorionic villi.

The procedure is always carried out under the guidance of an ultrasound scan to avoid making a hole in the amniotic sac (the protective sac that cushions the baby with fluid as it grows).

Potential talking points;

- “Would be interesting to know his point of view on having children due to the 50/50 chance of passing the condition onto his kids.”
- “Whether the disorder was life limiting and how this made you feel.”
- “I would have liked to know whether you have experienced any bullying or social exclusion. How you have dealt with this and what support you received.”
- “I think the only thing I would have liked to have known is what sort of impact psychologically and socially does OI have and has had on your life, and what sort of things can health professionals, charities or organisations do to make things easier in the future.”
- “Maybe include more of how we as nurses can make any stay you have in hospital better for you.”

TALK ON “Will there be a cure for OI?” if there is interest!