Seeing the ups when faced with downs - the complex genetic phenomenon of Down's syndrome and its impact in our society.

Introduction

In 2001, Karen Gaffney completed a relay swim of the English Channel. Six years later she swam the nine-mile span of Lake Tahoe.

Pablo Pineda, a Spanish actor completed his university degree in 2004 and went on to receive the Concha de Plata Award at the 2009 San Sebastián International Film Festival. He now works as a teacher.

Australian supermodel Madeline Stuart has appeared at New York, London and Paris fashion week and been featured in Vogue and Cosmopolitan after losing 20 kilograms weight to achieve her dream.

What do these three extraordinary people have in common? Namely a genetic condition called Down's syndrome.

Before the 1980s people with Down's syndrome were put into institutions, without adequate attention, education or medical care. They were neglected and essentially shunned from society for being 'no good'. A quote from a leading academic, American professor Joseph Fletcher in the 1960s; *"People [...] have no reason to feel guilty about putting a Down's syndrome baby away, whether it's "put away" in the sense of hidden in a sanatorium or in a more responsible lethal sense. It is sad; yes. Dreadful. But it carries no guilt. True guilt arises only from an offence against a person, and a Down's is not a person."* [FLETCHER 68]. This rather shocking statement was nothing strange at the time, but it just demonstrates the change in view of our society since then. I found this quote referenced in the book 'Far from the Tree' by Andrew Solomon. He uses this quote to highlight the view people had on those with Down's syndrome in history. Throughout his book he discusses what it means to bring up a child with a disability and offers a new perspective on life. Since 1983, the life expectancy for people with DS has increased by 35 years, from 25 to 60. Currently the oldest person alive with Down's syndrome in the UK is Georgie Wildgust, who turned 77 last year. Since the turn into the 21st century more and more people with Down's syndrome attend mainstream schools, more have graduated from high schools in the United States and more are going on to higher education. This change is recent, because of support and ambition, which has come about through a change in attitude.

It really has not been all that long for this large social change to have been made, and it just shows that once people see the reality, they stop focusing on what they consider perfection and learn to accept, support and enjoy. I am going to investigate the various aspects of Down's syndrome, looking into not only the genetics but also looking at its influence on life, for that of the person with Down's syndrome and those around them. I hope to offer historical, biological and social understanding complete with some personal views and prospective. I want to understand why and show that despite the limitations of Down's syndrome, people with the condition can lead semi-independent, fulfilled and happy lives, all whist improving society as a whole, by giving valuable lessons to those around them.

What is Down's Syndrome?

Down's Syndrome (DS) is a genetic condition which entails a set of physical, mental, and functional abnormalities as a result of an extra copy of chromosome 21 in the human genome, making 3 instead of the usual pair of two. This occurs during the creation of human sex cells (gametes) through meiotic division. It is a phenomenon known as trisomy 21, and it occurs in around 1 in 1,000 live births worldwide [THOMAS, 2019].

Down's Syndrome was officially recognised, and its characteristics described by John Langdon Down in 1866. However, it was not until 1958, some 92 years later that paediatrician and geneticist Jérôme Lejeune was able to find a genetic explanation for the condition. During a study of chromosomes of a child, he discovered the existence of an extra chromosome on the 21st pair. This was a significant milestone in genetics, as it was the first time in history a link was established between a mental disability and a chromosomal anomaly. Up until this point the condition was largely known as 'mongolism', after Langdon Down thought that cases of DS were a spontaneous step back in evolution from the supposed superior Caucasian race to the inferior Oriental race. This idea is absurd to us now, as well as extremely racist, and altogether a false representation of DS. The condition became known as trisomy 21, and increasingly over time called Down's Syndrome.

Since Lejeune's discovery, it has been shown that the characteristics and traits of DS are related to a small part of the long arm of chromosome 21, (21q22.1-21q22.3). This means the genetic material causing the effects is found between the first and third parts of the 22nd segment of chromosome 21. This small sub-section accounts for around 50-100 genes, which is a very small proportion of the whole human genome of around 30,000 genes [NEWTON, 2004]. All the genes of a human are contained within chromosomes, of which each person has 23 pairs in each cell, making 46 chromosomes. A person with DS will have 47.

Scientific research has shown that there are genetically three ways DS can occur with regards to chromosomal change. It is not a condition that is inherited, it is merely down to a chance error in cell division and can be diagnosed via blood tests. There are two different types of cell division within the body, mitosis and meiosis. Mitosis is used for most of the body's needs, whether it be replacing old and worn out cells, or adding new cells during growth and development. It results in two daughter cells which are genetically identical to the mother cell and contains the same number of chromosomes (46), making them diploid cells. Meiosis, however, is a type of bodily cell division which has only one purpose which is to produce sex cells, known as gametes. The male gamete is a sperm and the female, an egg. It results in four daughter cells, as opposed to the two in mitosis. These four daughter cells display genetic variation and have half the number of chromosomes as the mother cell (23), making them haploid cells. This means that when the male and female gametes fuse during fertilisation, a zygote with the usual 46 chromosomes is created, a diploid. Meiosis happens in two parts, meiosis I and meiosis II. Genetic variation during meiosis is brought about by crossing over and independent assortment of homologous pairs during the metaphase of meiosis I.

The most common way DS is caused is called non-disjunction or complete trisomy 21, and it occurs during the formation of either a sperm or an egg in meiosis. The extra chromosome usually comes from the mother, and in less than 5% of cases of non-disjunction does it come from the father. Non-disjunction is the failure of homologous chromosomes to separate properly during meiosis. If a cell divides and it results in more chromosomal material going to one pole than the other, then the daughter cells produced will have an abnormal number of chromosomes. In the case of trisomy 21, when the cell divides, both chromosomes of the homologous pair are brought to one pole in a failure to separate, rather than one to each. This means that one of the daughter gametes produced will have 24 chromosomes, with two copies of chromosome 21. The other will of course have 22 chromosomes, and no copies of chromosome 21. When the gamete with 24 chromosomes, whether it be a sperm or an egg fuses with its opposite-sex counterpart, a zygote with 47 overall chromosomes will be created, 24 from the abnormal gamete, and 23 from the other. A usual zygote will have two copies of each chromosome, however in trisomy 21 there is an extra copy of chromosome 21. When the zygote divides by mitosis and the rest of the body's cells are created, they too will display this extra chromosome.

Another cause of DS is translocation. It accounts for between 3-4% of all cases of DS. Translocation occurs during meiosis when a chromosome breaks off and attaches to another chromosome. This means that during the metaphase of meiosis I, an extra chromosome is brought to one pole instead of the other, connected to one of the other 23 chromosomes.

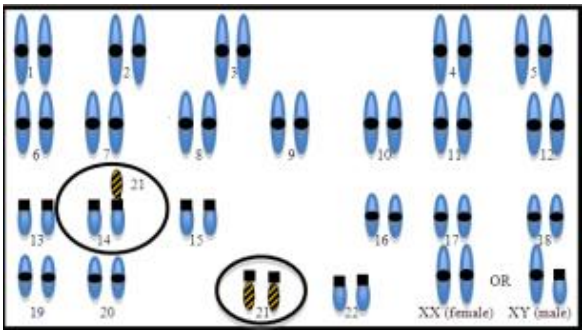

Fig. 1 **a diagram of the human genome with translocation of the 21st chromosome, attached to chromosome 14**

The chromosome which causes the features of DS is again the 21st which can be attached to chromosomes 13, 14, 15, 22, or in fact another chromosome 21. Few studies have been done to investigate the differences between those with translocation DS and non-disjunction, but they indicate that the differences are marginal, and they share essentially the same features [PRASHER, 1995]. The findings of certain studies, although requiring repetition with larger sample sizes do highlight an important area of further research. It is also possible, depending on how the chromosomes segregate on the equator of the spindle during meiosis, for the translocated material to go to the side from which it broke off attached to one of those 22 chromosomes. This is called balanced translocation, and the person will not display features of DS, as the correct amount of genetic information is present in the cell.

The third, and least common cause of DS is mosaicism. Mosaicism is a biological term which describes those who have two or more genetically different sets of cells in their body, meaning the chromosomal number within cells varies. While the other two causes were from errors during meiosis, mosaicism is from an error in mitotic cell division. As explained earlier mitosis is the body's main way for cells to divide, producing two diploid daughter cells that contain the same amount of genetic information as the parent cell. When an error occurs in mitosis the cell does not divide evenly into two. The result is that some cells contain 46

chromosomes, the normal 23 pairs, while others contain 47 or 45. The cells that then divide from the abnormal ones will also have that number of chromosomes. In the cases that cause DS, some cells will display trisomy 21 while others are normal. It occurs in one of two ways: either the zygote at first has three 21st chromosomes, which normally would result in simple trisomy 21, but during the course of cell division one or more cell lines loses one of the 21st chromosomes in a mitotic division error. The other way is vice versa; the initial zygote has two 21st chromosomes, but during the course of cell division one of the 21st chromosomes is duplicated. People with mosaic DS might have fewer characteristics of the condition than those with non-disjunction or translocation, if many at all; there is a variation. There has not been extensive research done to compare the two, however there was a resport published in 1991 by K Fishler and R Koch which talked about mental development in Down syndrome mosaicism. It compared 30 children with mosaic Down syndrome with 30 children with typical trisomy 21. After IQ tests, it was shown that the mean IQ of the mosaic group was 12 points higher than the mean of the non-mosaic group. Despite this, some children with typical Down syndrome did score higher on the IQ tests than some of the children with mosaic Down syndrome, highligting the variation. There has been an ongoing study project by the Department of Human Genetics at the Medical College of Virginia on children with mosaic DS. In a survey of 45 children with mosaicism, they found that these children did show delayed development compared to their siblings. 28 of these children with mosaicism DS were matched up with 28 children with typical DS for age and gender, and the children with mosaicism reached certain motor milestones earlier than children with typical DS, such as crawling and walking. However, speech development was equally delayed in both groups. It is certainly difficult to draw generalisations when the abilities of people with DS is so varied, but opens up possibility for further investigation.

DS is not a condition that is inherited, unless one of the parents of those with translocation DS themselves had a balanced translocation. In this case the parent is said to be a carrier of translocation DS and display no characteristics, however the translocation gene can be passed on to the child. Other than this genetic factor, the only other real factor that increases the risk of having a child with DS is advancing maternal age. Older mothers have a higher individual chance of having a baby with Down syndrome. At age 45, the risk of a having a child with Down syndrome increases to 1/30. [NEWBURGER, 2000]. Research has been done to investigate the reasons for this, through studies involviong mice. There are proteins present within cells that help keep chromosomes together at their centers. Lower levels of these proteins – called cohesin and securin – cause the chromosome pairs to be more loosely connected and futher apart. In a study by Nabti, Ibtissem et. al, it was found that older female mice had lower amounts of

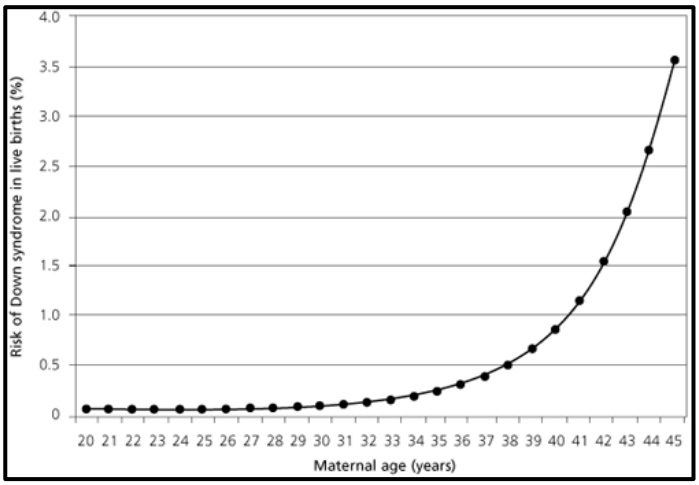

Fig. 2 **a graph illustrating the effect of maternal age on risk of conceiving a child with Down's Syndrome**

these proteins in their eggs, suggesting that as the eggs age, the levels of these proteins fall. This can then be related to human mothers, implying that the causes of DS previously described are more likely to occur as a result.

DS entails a set of very similar physical and mental characteristics for those who have it, albeit with some variation. Few people exhibit all the features, and some only exhibit a few. Some physical characteristics include shortness, a small head and ears, short neck, flat facial features, bulging tongue, upward slanting eyes, atypically shaped ears and low muscle tone. They tend to have more flexible joints, drier skin and sparser hair. The most consistent features are the facial appearance, skeletal structure leading to shorter stature and developmental anomalies of the heart. Those with DS are also more likely to develop visual and aural impairments and are at an increased risk of leukaemia and Alzheimer's disease. [NIA, 2017]. People with DS have an increased risk of cardiac disorders, most of which are congenital i.e. present from birth. Congenital cardiac disorders are present in around 40-60 percent of babies born with DS [PASCALL, 2015]. These are mainly septal defects, where part of the heart has not developed properly resulting in a hole being present. The most common congenital cardiac disorder in people with DS is an atrioventricular septal defect (AVSD). It occurs when the septum (partition) between the two ventricles of the heart and between the two atria of the heart does not develop properly and a hole is present. This means blood can mix from the left side to the right side of the heart. As a result, deoxygenated and oxygenated blood mix so the blood circulating the body has a lower level of oxygen, causing further health problems including Eisenmenger syndrome. Many of these heart defects require surgery very early on in the child's life, and until recently this was not known, hence why much fewer infants with DS survived beyond a few years of age. Because of their prevalence, screening for these issues must happen at a very young age. These problems, whether immediately life threatening or not, must be medically reviewed consistently.

Individuals with DS notably have behavioural and mental features as a result of their genes. These learning disabilities mean that it takes longer for them to develop certain skills, however level of ability certainly varies from person to person. Some typical behavioural attributes to DS can include a short attention span, poor judgment, and impulsive behaviour. Notable mental characteristics are delayed speech and language development, together with slow learning. What makes learning slower is a combination of factors including difficulties with receptive language and short-term memory, together with expressive delays. This means that academic, social and personal skills take longer for people with DS to develop, and most will never entirely reach that of someone without DS. These learning difficulties can be explained by the effect the extra chromosome has on the development of the brain. At a macroscopic level, variations can be seen in certain brain pathways. An example of this is the anterior commissure, a band of nerve cells connecting the two temporal lobes of the cerebral hemispheres across the midline of the brain, which are important for processing visual information and memory (see Figure 3). These appear smaller in those with DS, due to underdevelopment. In addition, the brain is made up of millions of nerve cells (neurones), which communicate with their neighbours through small outgrowths known as dendrites. The number and quality of these dendrites is what determines the level of processing and learning ability of the brain. In a person with DS, abnormal dendritic processes can be seen at

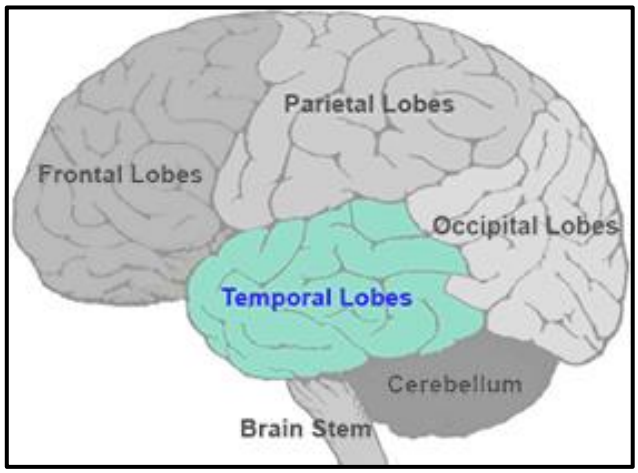

Fig 3: **diagram highlighting the temporal lobes of the brain**

a microscopic level. [NEWTON, 2004]. Despite the differences of the brains of those with DS, in the past the impairment of learning ability has been overemphasised, and full potentials have only been realised in recent years due to an access to adequate education, and widespread societal acceptance. It is certainly true that people with DS ted to have specific strengths regarding learning. It is said they have a distinct preference for visual learning, tending to be quite sharp with visual processing. They also tend to have high capacity for empathy and social understanding. They offer more comfort and support in times of distress in others. [KASARI, 2003]

The genes of those with DS are the same as everyone else's, just with an extra 1%. This is a very small amount in comparison to the whole human genome, meaning that all the rest of the genetic information is the same as it would be if the person did not have DS. [KESSLING, SAWTELL, 2002]. This extra information gives the potential rise to characteristics of DS, hence why features of those with DS are similar. However, as genes are inherited from parents, people with the condition look more like their family than others with DS.

Given the correct support and education, as it has been shown in recent years, it is certainly possible for people with DS to achieve personal goals and develop sufficient academic and communication skills to live semi-independent lives as they grow up, with full time jobs.

Life with Down's syndrome

Before I address life with DS, I would like to reference a short essay called "Welcome to Holland", written in 1987 by American author and social activist Emily Kingsley about having a child with a disability. It can be found in my references. [KINGSLEY, 1987]. This essay does not encapsulate all that it means to be a parent to a child with a disability, it merely provides a personal and more positive outlook. It shows that it certainly changes one's life, but that does not necessarily mean it changes it for the worse. Kingsley was mother to son Jason, born in 1974 with DS. She and her husband made a real effort to bring up their son in the best way possible, working hard on developing his skills. Jason became the first ever person with DS to appear on television, when he began to feature as a regular guest on *Sesame Street.* This was a big step as it normalised tolerance for a new generation, when he played with other children in a way that acknowledged but did not stigmatise his condition.

People with DS are known for being particularly affectionate and happy, with an excellent outlook on life. In a study approved by the Institutional Review Board of Boston University Medical Centre, 284 people with DS on the mailing lists of six non-profit Down's syndrome organisations around the United States were given a questionnaire on the subject of their lives and wellbeing. Those being surveyed were 12 and older. Among those surveyed,

almost 99% of people with DS indicated that they were happy with their lives; 97% liked who they are; and 96% liked how they look. Nearly 99% people with DS expressed love for their families, and 97% liked their siblings. These are some quite heart-warming statistics, and really demonstrate the mindset of those with DS, contented and compassionate and able to really develop loving and special relationships with those around them. As mentioned earlier, those with DS have their own relative strengths. Over time, most teens and adults are competent with self-help and daily living skills which often allows them to lead semi-independent lives, sometimes living by themselves or with roommates. With correct support and ambition, those with DS can strive for personal achievements such as being able to ride a bike, cooking their own meals, going to college, getting a driver's licence, getting married and having a full-time job. Things for people with DS are often done at a slower pace, with more time needed and potentially with a slightly different approach, but this does not make it all impossible. Relating back to the achievements I mentioned at the start, it just shows what is possible.

With regards to the preparation for life – I have mentioned that things are often done slightly differently for people with DS, more support is needed, and they perhaps have different goals to others. While this is true, it is still very important for theirs and others sense of inclusion and acceptance that they are treated like anyone else, with the same opportunity and freedom. A largescale example of this is with schooling, although all students with DS considerably benefit from having personal learning assistant or a one-to-one. A survey was completed in 1999 which concluded that teenagers with DS educated in mainstream schools are gaining considerable benefits in academic skills, communication skills and social independence [BUCKLEY, BIRD 99]. It was a survey of the progress of 46 teenagers with DS in Hampshire, of whom 18 are in mainstream secondary education and 28 are in special schools. Information was collected on a wide range of issues including, health, behaviour, sexuality and social lives in addition to personal and social independence, communication skills and academic progress. The general trend showed that on average those with DS from a mainstream school were more competent with these tasks than those from the special schools. The surveys were carried out using questionnaires for the parents – the Sacks & Buckley Questionnaire (SBQ) and the Vineland Adaptive Behaviour Scale (VABS). With regards to communication, it was shown that the expressive language of those from the mainstream schools was 2 years and 6 months ahead of the special school group. Some 78% of the mainstream teenagers are rated as being intelligible to strangers compared with 56% in special schools in 1999. This demonstrates that the atmosphere and nurture of mainstream schools provides those students with DS appropriate and more developed communicative skills. They are very inclusive places, those with DS mix with those without and it creates a more equal, rather than separated school community. The other classmates also learn a great amount too, and it provides a valuable and eye-opening experience for everyone else within that school. In addition, the significance of reported instances of difficult behaviour of the children was lower for those from the mainstream schools than the special schools (see Figure 4). It can be seen that difficult behaviour affects the learning and social opportunities of a teenager with DS and can create stress for teachers and families.

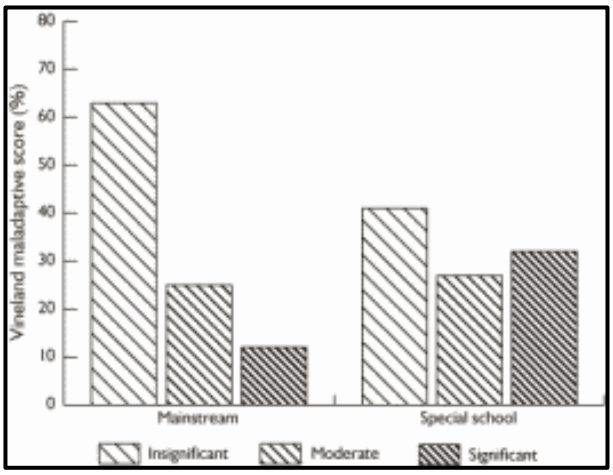

Fig. 4: **The significance of reported behaviour difficulties (percentage of teenagers in each category) [BUCKLEY, BIRD 99]**

Contrarily, teenagers who can behave in a socially acceptable and competent manner will be more likely to have friends, have active social lives and to be successful in work as adults.

Some comments on the positive aspects of the teenagers personalities were made from parents involved in the study: “He is popular, friendly and non-judgemental… he has added another dimension to our lives." "Our daughter brings more love, fun and laughter to family life and though she will never be 'academic' there are other qualities she has which cannot be measured." These comments show first-hand the impact these children have on the people around them.

As shown here with these quotes, there is no doubt that there is a truly special benefit these people with DS give to life for those around them. In a large-scale, population-based study, significantly lower divorce rates were found among families of children with Down syndrome (7.6%) compared with those of children with no identified disability (11.2%). The study involved the families of 647 children with Down syndrome and 361,154 families of comparison children who did not have any record of congenital birth defects (the comparison group). [URBANO, HODAPP 07] This is a remarkable statistic as it shows that something like a disability does not break families apart, but instead seems to bring them closer together. It links quite nicely with the study mentioned before about wellbeing, regarding the statistics about liking family and siblings. It encapsulates the connection within the family and shows that all the things the child brings to the family are valued and cherished.

I was able to contact some parents of children with DS who are part of a local charity my family are involved in via email, in order to gauge more about their view on how having a child with DS has impacted their life. I questioned them on the most valuable thing they had learned from having their child. “Priorities”, “Appreciation” and “Love” was what trustee Tatty Bowman told me were the three most important things she has been lucky enough to consider and develop after having and bringing up her son George. She has “no doubt he has made [her] a better person”. “It brings a blessed view of life” told Emma Eichhorn, mother to Henry, on the perspective it has given her. Ellie Martin mentioned that the biggest thing she and her family have learned is the “power of acceptance”, and the value of embracing what life gives them. (for full responses see Appendix). These truly honest and powerful words I was met with really expresses the eye-opening viewpoint of experiencing first-hand something that is completely different to what people are used to, or what they expected. From this I can learn that despite the challenges raising a child with DS may pose, the initial dread is soon met and replaced by such wonderful qualities, which ultimately lead to a better view on life.

Conclusion

When I was 5 years old, my younger sister, Lia, was born and diagnosed with Down's syndrome. Naïve and confused, I did not really know what to make of it, and the idea that my sister was not going to be normal was a complex thought. Until this point, whilst I was aware of disability in the world, I was seemingly ignorant. I was still like this for years to come but as I grew, I learned to understand it all more since I now had the experience in my own household. Life was different at times, yes, and I had many things to learn. Life is spontaneous and changes in an instant but being able to work with what life gives you, figure out new paths and plans is a truly enriching and rewarding experience. It has allowed me to put everything into perspective, appreciate the small things in life and really learn to accept, support and enjoy. While Lia (now 11) is a little different to others her age in some ways, most ways she is just like any other child. She is funny, ambitious, curious and just as annoying as my older sister. I have shared some of my best quality moments with her as we share the same sense of humour and watching her take pride in her own achievements and activities is incredibly special.

I have been curious and fascinated my whole life about this condition, and this project has been an opportunity to really develop my understanding, and research further into the various different aspects of DS. I watched a TED talk performed by Heather Lanier, mother to daughter Fiona who has Wolf-Hirschhorn syndrome; a genetic condition that results in developmental delays. '"Good" and "bad" are incomplete stories we tell ourselves' was the title. She talks about what she has learned from having her daughter. She says Fiona has "rare blueprints" and is not designed to be like other people. Life is not factory perfect and genetic flaws are inevitable, and most importantly very much part of our world. She was able to drop her misconceptions on what was "good" or "bad", challenging us to stop fixating on solutions for whatever we deem not normal, and instead to take life as it comes. It can be applied for all people with a disability. They are different, yes, but does that mean bad?

I have learned that there are undoubtedly limitations for those with DS and they will almost certainly not have the level of academic or social skills as their peers, and they may find it harder to develop these skills, but that does not mean that it is not worth the effort. In addition, I have learned about the complexities of the extra chromosome, and what effect it has physically and mentally. It is all accounted for by an extra 1% of genetic information, and the rest is all passed on by the parents just like any other child. This has shown me that indeed there are different characteristics and physical and mental difficulties associated with DS, but that does not make people who have DS any less human, and indeed with their own strengths and support from others, they can lead their own semi-independent lives. Countless successful people with DS can be found all over the internet and there is much evidence for society's changing views. Their condition does not prevent them from getting the most out of life, inspiring others and being a positive and beneficial part of society – and they seem to have a remarkable ability to make people smile. In fact, the world learns a lot from these individuals, which is often overlooked. The same could apply to all people with disabilities, mental and physical. When we learn to accept and understand difference, we learn to value it, rather than judge or criticise. I have learned that everything people with DS bring to this world; all the lessons, love, enjoyment, determination and attitude, makes their life every bit as worth living as mine or yours.

Figure 5**. Sienna (left), Lia (centre), Theo (right) at Bodiam Castle**

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Kingsley, Emily Pearl (cited 4/7/20) (1987) 'Welcome to Holland' I am often asked to describe the experience of raising a child with a disability - to try to help people who have not shared that unique experience to understand it, to imagine how it would feel. It's like this.....

When you're going to have a baby, it's like planning a fabulous vacation trip - to Italy. You buy a bunch of guidebooks and make your wonderful plans. The Coliseum. The Michelangelo David. The gondolas in Venice. You may learn some handy phrases in Italian. It's all very exciting.

After months of eager anticipation, the day finally arrives. You pack your bags and off you go. Several hours later, the plane lands. The stewardess comes in and says, "Welcome to Holland."

"Holland?!?" you say. "What do you mean Holland?? I signed up for Italy! I'm supposed to be in Italy. All my life I've dreamed of going to Italy."

But there's been a change in the flight plan. They've landed in Holland and there you must stay.

The important thing is that they haven't taken you to a horrible, disgusting, filthy place, full of pestilence, famine and disease. It's just a different place.

So, you must go out and buy new guidebooks. And you must learn a whole new language. And you will meet a whole new group of people you would never have met.

It's just a different place. It's slower paced than Italy, less flashy than Italy. But after you've been there for a while and you catch your breath, you look around.... and you begin to notice that Holland has windmills....and Holland has tulips. Holland even has Rembrandts.

But everyone you know is busy coming and going from Italy... and they're all bragging about what a wonderful time they had there. And for the rest of your life, you will say "Yes, that's where I was supposed to go. That's what I had planned."

And the pain of that will never, ever, ever, ever go away... because the loss of that dream is a very, very significant loss.
But... if you spend your life mourning the fact that you didn't get to Italy, you may never be free to enjoy the very special, the very lovely things ... about Holland.

Figures

Figure 1: diagram of the human genome with translocation of the 21st chromosome, attached to chromosome 14 (cited 10/5/20) (reviewed 2020) University of Rochester Medical centre https://www.urmc.rochester.edu/encyclopedia/content.aspx?contenttypeid=90&contentid=P02153

Figure 2: graph illustrating the effect of maternal age on risk of conceiving a child with Down's Syndrome by Newburger, David S M.D., State University of New York at Buffalo, Buffalo, New York 2000 Aug 15;62(4):825-832. (cited 16/5/20) (2000) Down Syndrome: Prenatal Risk Assessment and Diagnosis – American Family Physician https://www.aafp.org/afp/2000/0815/p825.html

Figure 3: diagram illustrating the lobes of the brain, as depicted by Queensland Health from the Queensland Government website (cited 22/5/20) (18 Apr 2017) https://www.health.qld.gov.au/abios/asp/btemporal_lobes

Figure 4: graph illustrating the significance of reported behaviour difficulties (percentage of teenagers in each category) from a study by BIRD, BUCKLEY 99. (cited 1/7/20) (2006) A comparison of mainstream and special education for teenagers with Down syndrome: Implications for parents and teachers. Down Syndrome Research and Practice, 9(3), 54-67. https://library.down-syndrome.org/en-gb/research-practice/09/3/comparison-mainstream-special-education-teenagers-down-syndrome-implications-parents-teachers/

Figure 5. Photo of Lia, Sienna and Theo at Bodiam Castle (cited 7/7/20) (2019) iPhone

Appendix

Questionnaire study to mothers of those with DS from local charity, email correspondence with myself, T Lakin.

Question posed: *'What is the most valuable thing you have learned from having and raising your child?'*

Responses:

"The biggest thing we have learned is the power of acceptance. The value of embracing what life has given us, navigating the fears and uncertainty and ultimately learning that there are some things in life we cannot control. It's led us to new ways to think about and appreciate things which has ultimately led to a much richer happiness." – ***Ellie Martin 30/6/20***

"Patience. Before I had L (now aged 8) I experienced my eldest son hitting development milestones, largely ahead of his NCT peer group. As a first time mum it pleased me that he was the first to sit, the first to try 'proper' food and he was babbling brilliantly and he was doing this without any input from me, it just happened. Then L came along and first I had to teach him to breast feed… every feed took over an hour, every step repeated for him every feed, he eventually learnt! And every step along the way I've had to break down a task, a task that D (eldest son) just did, into small, incremental steps and teach it to L. Each step being patient, not getting frustrated, repeating it over and over until he gets it. And he does. That's it. He gets there. In his own time. He has taught me that it's fine to take things slower, it doesn't matter how long it takes so long as we keep trying. And those milestones that D, and then his younger brother C, sailed to and flew past, L is achieving in the time scale that works for him. And that's perfectly great by me. I'm patient. And it's allowed me to be patient with life in general." - ***Karen McGuigan 30/6/20***

"Can I tell you three things I have learned?

Priorities. I have learned that an awful lot of what we worry about really doesn't matter. We waste much energy on fretting about this and that when compared to the big things, they are of no consequence. So, to use a well-known quote I "don't fret the small stuff" anymore!

Appreciation. I have learned to appreciate every day. Life can change at the drop of a hat and nothing is guaranteed. So, when someone you love walks into a room smile at them, when you have an opportunity be kind to people, because you never know what tomorrow holds.

Love. You can love anyone. I have learned that the love I feel for G is fierce. Without ever being able to tell me he loves me too (he has no words or communication system) I have fallen so hard for this contrary, tricky, gorgeous boy. So, love really does conquer all!!!!" – ***Tatty Bowman 30/6/20***

"We love that R truly exemplifies the term" neurodiversity." She doesn't see the world, or comment about the world in the way that "the majority would". To illustrate this...Last week she got new sunglasses. She put them on but happened to have her back to her father. Tim said to her "R - can I see your new glasses." She took them off and handed them to him! She took him very literally and missed the unspoken nuance of "let me see how good you look in your new glasses." My predictions is that "most people would have turned to show their father their glasses and kept them on their face. I love that R does it differently - not wrong - but not "classic.": Neurodiversity is so enriching but we often go through life too fast to notice it or appreciate it and we miss the smile it brings when we see the world through a different lens (no pun intended!)" – ***Sarah Hodgson 30/6/20***

"I am mum to H who is 9 and I would say the most valuable thing I've learned through having him in our lives is perspective. H was born with tummy issues and has been through many tests and surgeries over the years, dealing with it all with such strength and determination. His learning needs were my biggest worry when he was born, but his health has given me a different, more precious view on life, that I do not think I would have had otherwise." – ***Emma Eichhorn 30/6/20***

"My answer would be that I have learnt to be patient and understanding of a person with special needs when trying to teach them and help them. Patience most of all. There is no good going too fast, asking too much, expecting too much and not giving your time. You get a lot more out of someone if you are patient and understanding of them. This of course transpires into everyday life and being patient with all people and understanding of the complexity of humans!" – ***Louise Beattie 1/7/20***

"So, what have we learned from J? To not take parenthood for granted. We are very lucky. We see life more simply and honestly. Jude has taught us to celebrate and appreciate the simple things in life." – ***Orna McCullough 6/7/20***