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The "Tragedy" of Down Syndrome: Is it Really So Tragic?

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THE "TRAGEDY" OF DOWN SYNDROME:

IS IT REALLY SO TRAGIC?

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Introduction

For many expectant parents, receiving the news that they are going to have a baby is a joyous and exciting period in their lives. It is the start of a journey that will permanently alter their lives, leading to an addition to their family unit. Over the next several months, expectant parents are consumed by thoughts of the impending birth, preparations for the new addition, and imagining what life will be like once the baby has arrived. Then, there are also thoughts of the future: baby's first word, baby's first day of school, baby's graduation, baby's wedding, and any other number of milestones. Somewhere among all of these thoughts are also the worries for every potential problem that could occur. What if something goes wrong with the pregnancy? What if the baby is born prematurely? What if the baby is born with a disorder? The possibilities are endless. Because of these fears, it is not unusual for a parent to express the desire for the baby to be born healthy with ten fingers and ten toes. Why would the parent not want his or her child to be healthy and "normal"?

Often these fears are unnecessary, but occasionally expectant or new parents receive the news that most parents dread: something is not quite right with their baby. Because it is our automatic thought to imagine a baby being healthy and perfect in every way, receiving this news comes as a shock, often incomparable to any other shock experienced. Even if the parents prepare themselves for the possibility of their child joining the statistics of being one in every certain number of children born with this disability or this feature, the shock still exists. Until a doctor confirms a diagnosis or gives that prognosis for the child's life, hope still remains that this is simply one of those bad dreams or cruel jokes.

Down syndrome is one of these diagnoses that most parents dread to hear from their doctors. Hopes and dreams come crashing down and new fears are piled on top of the existing worries that accompany the baby's birth. According to the National Down Syndrome Society

(2012), "one in every 691 babies in the United States is born with Down syndrome, making Down syndrome the most common genetic condition". However, each year in the United States 3,999,386 babies are born (Centers for Disease Control and Prevention, 2012). Comparing these two figures and seeing the vast difference in the totals, it can be better understood why a parent would receive such a shock when hearing that their precious bundle of joy has Down syndrome.

For most families, the birth of a child is often considered to be a "miracle" or a "blessing." The joy and warmth surrounding the newborn is contagious and one of a parent's most treasured memories. However, the mood surrounding the birth of a child with Down syndrome is often much different. Fear, anger, shock, worry, and a number of other emotions can plague the arrival of the newborn who is diagnosed with Down syndrome. Words such as "miracle" and "blessing" may not accompany such an arrival. More often than not, the delivery of a child with Down syndrome is considered to be a "tragedy": both for what has been lost and what could have been.

In our society, we strive for perfection and normalcy. Children with Down syndrome do not easily fit into this "perfect" world that we seem to believe exists. Because of a chromosomal error that they had no control over, children with Down syndrome can be viewed at first as less desirable in comparison to the image the parent had in mind. Many factors contribute to this illusion of Down syndrome being a tragedy. One factor is the general publics' limited knowledge of the diagnosis, which can sometimes be full of misconceptions. These ideas or lack thereof can impact how a newborn baby with Down syndrome is welcomed into this world. Another contributing factor to the illusion that Down syndrome is a tragedy is the way that the diagnosis is presented to families by medical professionals. If a doctor presents the idea that a child will never be a normal functioning individual or seems to strongly lean toward terminating the

pregnancy following a prenatal diagnosis of Down syndrome, the parents are more often than not going to believe what the doctor is presenting to them. Unfortunately, some medical professionals also seem to fall into the category of a misinformed society, and false information provided by them is influencing parents. Another factor that enables people to believe in the tragedy of the disorder is the quality of support presented to families following a diagnosis of Down syndrome. If little support is provided and families feel alone in the situation, then it is no wonder why many of them view the situation as tragic.

All of these factors prove that something needs to be done to educate the general public and medical professionals on the topic of Down syndrome. Because of this misinformation, Down syndrome is not being presented as simply part of the child but rather as the child's identity. Following this diagnosis, many parents question whether or not they would be able to raise a child with Down syndrome and are being forced to decide whether it is best to keep the child, give the newborn up for adoption, or terminate the pregnancy. Such drastic measures result from misinformation. These common misconceptions that individuals hold could easily be addressed if our society could be informed on what Down syndrome is, the prenatal and postnatal assessments available to families in determining Down syndrome, and the resources available to parents prior to or following a diagnosis of Down syndrome. The purpose of this thesis is to show that a diagnosis of Down syndrome should not be considered a tragedy. By reviewing relevant literature, this thesis will provide readers with accurate information on Down syndrome and discuss the implications of misinformation as well as the lack of information provided to parents. This will result in a recommendation for more training for medical professionals as well as sufficient support for every family.

Literature Review

Down syndrome

While Down syndrome is the most common genetic condition, many people do not know all of the facts surrounding the diagnosis. In the most basic of terms, Down syndrome is a disorder that affects the physical, social, and intellectual development of an individual. The human body consists of trillions of cells; and within each cell, there is a nucleus, which contains the genetic material for the cell. This genetic material is housed in genes, which "carry the codes responsible for all of our inherited traits and are grouped along rod-like structures called chromosomes" (National Down Syndrome Society, 2012). While most people have 23 pairs of chromosomes, people with Down syndrome have a "chromosomal abnormality" that results in either an entire extra copy or an extra portion of the 21st chromosome, which affects the individual's development (Roizen, 2007, p. 263).

Unlike some disorders, Down syndrome can occur in both genders, as well as in any race, nationality, religion, or socioeconomic status (National Association for Down Syndrome, 2012). It can affect any family; however, women over the age of thirty-five have a much higher risk of carrying a child with Down syndrome. A mother who is twenty-five years of age has a 1 in 1,200 chance of conceiving a child with Down syndrome, while a mother who is thirty-five years of age has a 1 in 350 chance of conceiving a child with Down syndrome. While the incidence increases with maternal age, it has been found that eighty percent of infants diagnosed with Down syndrome are born to mothers who are under thirty-five years of age (National Association for Down Syndrome, 2012). Below, Figure 1 shows the incidence of Down syndrome based upon the maternal age. In each column, maternal age is listed on the left, and on

the right, is the incidence of Down syndrome. The table shows how the incidence of Down syndrome increases as maternal age increases.

Maternal Age	Incidence of Down syndrome	Maternal Age	Incidence of Down syndrome	Maternal Age	Incidence of Down syndrome
20	1 in 2,000	30	1 in 900	40	1 in 100
21	1 in 1,700	31	1 in 800	41	1 in 80
22	1 in 1,500	32	1 in 720	42	1 in 70
23	1 in 1,400	33	1 in 600	43	1 in 50
24	1 in 1,300	34	1 in 150	44	1 in 40
25	1 in 1,200	35	1 in 350	45	1 in 30
26	1 in 1,100	36	1 in 300	46	1 in 25
27	1 in 1,050	37	1 in 250	47	1 in 20
28	1 in 1,000	38	1 in 200	48	1 in 15
29	1 in 950	39	1 in 150	49	1 in 10

Figure 1, Incidence of Down syndrome based upon Maternal Age
http://www.ndss.org/Down-Syndrome/What-Is-Down-Syndrome/

There are three types of Down syndrome, but each type affects chromosome 21 in some way. The most common type of Down syndrome is trisomy 21and affects ninety-five percent of individuals diagnosed with Down syndrome (Roizen, 2007, p. 263). Trisomy 21 results from nondisjunction, which is when an error occurs as the cell divides. In Down syndrome, this "results in an embryo with three copies of chromosome 21 instead of the usual two" (National Down Syndrome Society, 2012). This error occurs prior to or at conception when "a pair of 21st chromosomes in either the sperm or the egg fails to separate" and is repeated in every cell within the body (National Down Syndrome Society, 2012). Below, Figure 2 depicts how a cell normally divides. Both the egg and the sperm carry 23 chromosomes, and when fertilization occurs, the cell receives 23 chromosomes from the egg and 23 chromosomes from the sperm. This results in 46 chromosomes in the cell. The cell divides, and the outcome is two cells with identical sets of chromosomes. This copying of the chromosomal pairs is continued each time as the cell divides.

trisomy 21, translocation results in the typical 46 chromosomes in each cell; however, a portion of chromosome 21 "breaks off during cell division and attaches to another chromosome, typically chromosome 14" (National Down Syndrome Society, 2012). While Down syndrome is not inherited in most cases, translocation accounts for the few cases when one of the parents may be a carrier (Boston Children's Hospital, 2013). This does not occur in all instances of translocation, however.

The final type of Down syndrome is Mosaicism, and it is the rarest of the three types. Only about 1 percent of individuals are diagnosed with mosaic Down syndrome (Roizen, 2007). Mosaic Down syndrome is also the result of nondisjunction of the 21st chromosome; however, this "takes place in one – but not all – of the initial cell divisions after fertilizations" (National Down Syndrome Society, 2012). This means that some cells contain the usual 46 chromosomes, while others contain 47 chromosomes. The cells with 47 chromosomes hold the additional material from the 21st chromosome. According to the National Association for Down Syndrome (2012), the cells appear in a mosaic pattern, which is where the term "mosaicism" stems. Figure 4 shows how some cells contain 46 chromosomes and others hold 47 chromosomes.

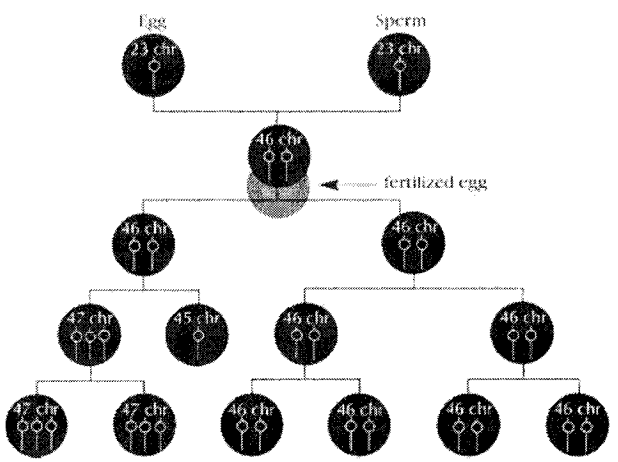

Figure 4, Mosaic Cell Division
http://www.ndss.org/Down-Syndrome/What-Is-Down-Syndrome/

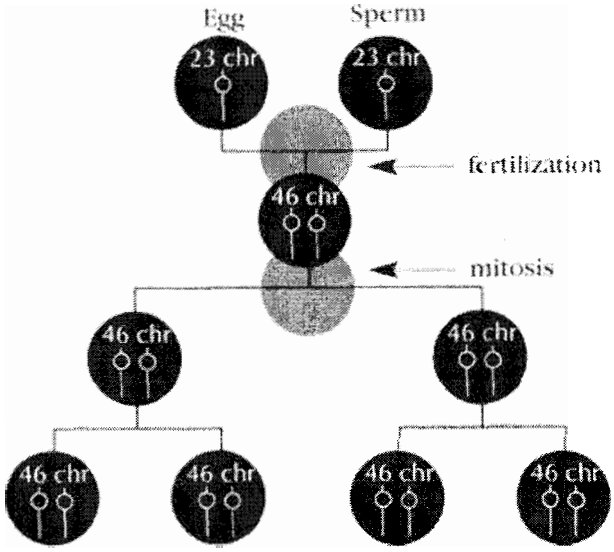

Figure 2, Typical Cell Division
http://www.ndss.org/Down-Syndrome/What-Is-Down-Syndrome/

Figure 3 shows how a cell divides when nondisjunction occurs, and the individual has three copies of chromosome 21. This results in 47 chromosomes in each of the cells, instead of the typical 46 chromosomes.

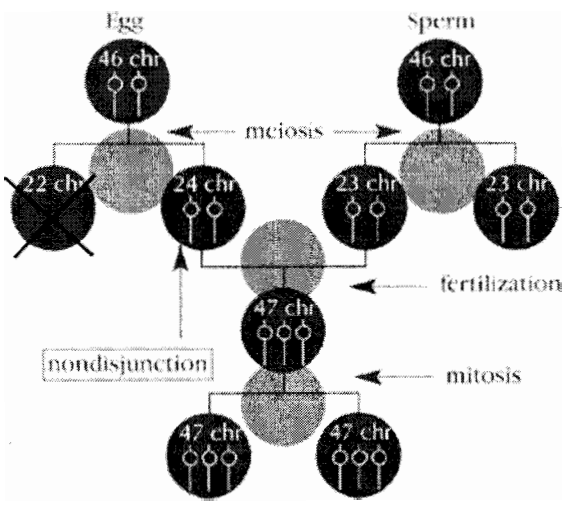

Figure 3, Trisomy 21 (Nondisjunction) Cell Division
http://www.ndss.org/Down-Syndrome/What-Is-Down-Syndrome/

Another type of Down syndrome is translocation, which occurs in approximately four percent of individuals who are diagnosed with the chromosomal disorder (Roizen, 2007). Unlike

trisomy 21, translocation results in the typical 46 chromosomes in each cell; however, a portion of chromosome 21 "breaks off during cell division and attaches to another chromosome, typically chromosome 14" (National Down Syndrome Society, 2012). While Down syndrome is not inherited in most cases, translocation accounts for the few cases when one of the parents may be a carrier (Boston Children's Hospital, 2013). This does not occur in all instances of translocation, however.

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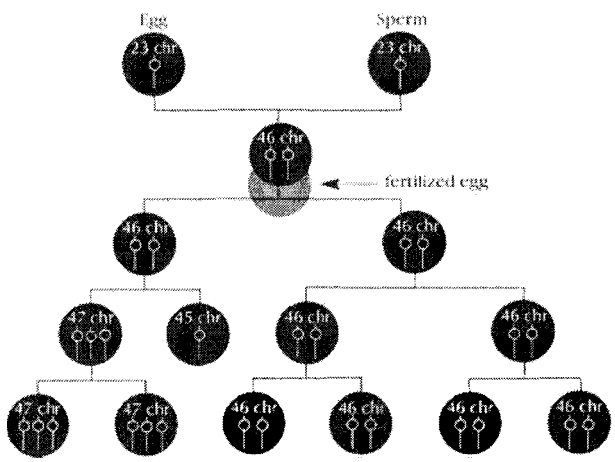

Figure 4, Mosaic Cell Division
http://www.ndss.org/Down-Syndrome/What-Is-Down-Syndrome/

There are several different physical characteristics that are common among individuals with Down syndrome. Some of these characteristics include "three palm print (dermatoglyphic) patterns, Brushfield spots (colored speckles in the iris of the eye), …neck skinfold, and widely spaced first toes" (Roizen, 2007, p. 264). Children with trisomy 21 and translocation share many of these characteristics, making it impossible to determine which type the child has simply by looking at him or her. Many of the physical features mentioned earlier are not as prominent in children with mosaic Down syndrome; this may attribute to their later diagnoses. According to the Down syndrome Association of Orange County (2013), the "average age for individuals to be diagnosed with Mosaic Down syndrome is 1-4 years of age". Some people receive this diagnosis much later in life. There are also several medical concerns and increased health risks associated with Down syndrome. Some more common concerns include congenital heart defects, ophthalmic disorders, hearing loss, and growth problems (Roizen, 2007). The list is quite extensive, and there is an "increased risk of abnormalities in almost every organ system" (Roizen, 2007, p. 264). However, as with every diagnosis, the presence of different physical characteristics and the severity of the various medical concerns differ for each child.

Diagnosing Down syndrome

Diagnosing a child with Down syndrome can be done in a few ways. One of those ways is through prenatal testing. With the many advances in technology over the years, prenatal testing has become a more common practice for many expectant parents. Prenatal testing provides parents with either the likelihood of their child having a disorder or the actual diagnosis of a disorder. Within prenatal testing, parents are given the option to undergo a screening test and potentially a diagnostic test. A screening test provides parents with the "risk" of their child being

born with Down syndrome, while a diagnostic test can give parents a more definite answer (National Association for Down syndrome, 2012). Screening tests are often less invasive than diagnostic testing; they typically require an ultrasound and a blood sample. This combined test is done within the first trimester and in two steps during week eleven to thirteen of the pregnancy. An ultrasound, known as the nuchal translucency screening test, is done to "measure a specific region on the back of a baby's neck" to see the amount of fluid present (Mayo Clinic, 2011). If there is an abnormality, "more fluid than usual tends to collect in this tissue" (Mayo Clinic, 2011). The next portion of the test is the blood samples, which are compared with the ultrasound to determine the possibility that the child will have Down syndrome (Mayo Clinic, 2011).

In previous years, prenatal testing has been recommended to women who are older and therefore more likely to carry a child with Down syndrome. However, today "prenatal screening tests are…routinely offered to women of all ages," and if the results indicate a higher probability of having a child with Down syndrome, "doctors will often advise a mother to undergo diagnostic testing" (National Down Syndrome Society, 2012). While it is beneficial to confirm a diagnosis of Down syndrome, diagnostic testing is much more invasive than screening tests and carries a higher risk of miscarriage. Because of these reasons, parents are more wary of performing one of these tests in order to receive that confirmation. A common diagnostic test that can confirm Down syndrome is an amniocentesis, which collects a "sample of the amniotic fluid surrounding the fetus…through a needle inserted into the mother's uterus" (Mayo Clinic, 2011). From the sample, the chromosomes of the fetus are analyzed to identify if the child has Down syndrome (Mayo Clinic, 2011). This test is typically performed after 15 weeks of

gestation, and the chances of spontaneously terminating the fetus are 1 in 200 (Mayo Clinic, 2011).

Another common form of testing to identify Down syndrome is the chorionic villus sampling (CVS). This test is typically performed between 9 and 14 weeks of gestation and "carries a 1 in 100 risk of miscarriage" (Mayo Clinic, 2011). During this test, cells are extracted from the placenta, and the chromosomes are analyzed to identify Down syndrome (Mayo Clinic, 2011). Both of these tests will answer parents' questions on whether or not their child has Down syndrome; however, because of the risks associated with the testing procedures, parents are encouraged to take some time deciding if they want to pursue this option.

Not all families receive a prenatal diagnosis of Down syndrome. Some parents are informed of the diagnosis following the birth of their son or daughter. Because Down syndrome has specific physical characteristics associated with its diagnosis, doctors are usually able to identify its presence at birth. Children will often have the physical traits mentioned earlier, such as the three palm print pattern, Brushfield spots, as well as upward slant of the eyes, or low muscle tone (National Down Syndrome Society, 2012). If a physician recognizes some or all of these features, he or she will have the child undergo a diagnostic test called a karyotype. A blood sample is collected from the infant, and the chromosomes are analyzed to determine if the child has Down syndrome (National Down Syndrome Society, 2012). Because the diagnosis is not always suspected prior to birth, many parents receive the news following the child's arrival.

Outcomes from Receiving a Diagnosis of Down syndrome

As mentioned earlier, there are a few different ways Down syndrome can be diagnosed. Numerous sources have arrived at the general conclusion that prenatal testing has been both

beneficial and detrimental to Down syndrome. Brian G. Skotko's (2005a) journal article "Prenatally Diagnosed Down Syndrome: Mothers Who Continued Their Pregnancies Evaluate Health Care Providers" reported the results from a survey he designed that was sent out to parents with a child or children with Down syndrome. Of the 2945 surveys mailed out, 1126 surveys were returned and 141 of those women were from mothers who had received prenatal testing. The survey showed that mothers who received a prenatal diagnosis and chose to continue the pregnancy were "generally happier over the birth of their infant with [Down syndrome] DS than their counterparts who had received the diagnosis postnatally" (Skotko, 2005a, p. 676). It was projected by Skotko (2005a) that this more positive emotion from those mothers who had received a prenatal diagnosis was due to the ability to "resolve any grief before their child was born" (p. 676). Essentially, they were more "prepared" for the different demands that arise when raising a child with Down syndrome.

Because there were only 141 mothers who received prenatal testing out of the 1126 surveys returned, Skotko (2005a) determined that "the majority of women who have fetuses with DS still find out about the diagnosis postnatally, or…a large number of women who receive prenatal diagnoses of DS choose to terminate their pregnancies" (p.675). He also considered a third option that both of these are occurring. Many individuals do not have an extensive knowledge of Down syndrome; therefore, a diagnosis of Down syndrome can seem very worrisome or even terrifying for some parents. In the article "Prenatal Testing for Down Syndrome: Comparison of Screening Practices in the UK and USA," there was a case study of a 37 year old American woman named Marianne, who was living in England. Marianne "felt quite concerned about the possibility of having a baby with Down Syndrome" (Tapon, 2010, p.118). She and her husband chose to scan for Down syndrome, and it was discovered that her baby

seemed normal except for one marker. They were informed that there was a low risk of their baby having Down syndrome, so they chose not to do more invasive testing to be sure. Following the birth of their child, it was determined that the baby did have Down syndrome. A genetic counselor spoke with Marianne and her husband about their options, and they "declined giving the baby for adoption stating they now felt committed to their baby" (Tapon, 2010, p. 119). They also expressed their "frustration with the screening test, stating that they would have terminated the pregnancy had they known the baby had Down syndrome" (Tapon, 2010, p.119). It is difficult to tell from the article the extent of the couple's knowledge regarding Down syndrome; however, it is clear that they had very strong beliefs on the subject. Like many parents, they felt that their child having a diagnosis of Down syndrome was a tragedy, and they would have ended the pregnancy had they known the final outcome.

According to the article "Prenatal Diagnosis of Down Syndrome: A Systematic Review of Termination Rates (1995-2011)," the "choice about whether or not to continue a pregnancy following a prenatal diagnosis of Down syndrome is a complex decision motivated by attitudes towards termination, socioeconomic factors, ultrasound findings, and other considerations" (Natoli, Ackerman, McDermott, & Edwards, 2012, p. 142). This choice has been presented to countless parents over the years, and rates of termination have remained quite high with more than half of the pregnancies being terminated. Unfortunately, there are few studies that have extensively gathered and examined termination rates following a diagnosis of Down syndrome. Taking a look at the Mansfield et al. review, "Termination Rates After Prenatal Diagnosis of Down Syndrome, Spina Bifida, Anencephaly, and Turner and Klinefelter Syndromes: A Systematic Literature Review," the literature review provides an estimation of termination rates following prenatal diagnoses across several countries over many years (Mansfield, Hopfer, &

Marteau, 1999). Based on responses from a variety of developed countries, it was determined that 92% of mothers would choose to terminate the pregnancy following a Down syndrome diagnosis (Mansfield et al., 1999). Because this study is one of very few regarding termination rates following a diagnosis of Down syndrome and is now somewhat outdated, Natoli et al., cited earlier in this review, decided to conduct a more recent survey.

In 2012, Natoli et al. reviewed different studies that reported data for pregnancies with prenatal diagnoses of Down syndrome, which were also terminated. The studies reviewed were placed into categories: "population-based studies (e.g. statewide birth defect registries)" and "hospital-based studies (e.g. single-institution studies)" (Natoli et al., 2012, p. 144). Seven population-based studies were reviewed, and it was determined that 67% of pregnancies following a prenatal diagnosis of Down syndrome were terminated (Natoli et al., 2012). Data from nine hospital-based surveys was also examined, and 85% of pregnancies were terminated once the parent was given a prenatal diagnosis of Down syndrome (Natoli et al., 2012). One limitation of this study, however, is that data was collected from only eight states within the United States and the District of Columbia. Data was collected from different regions of the United States, but the termination rates may be different if data was used from more states. Although the termination rates of 67%-85% reported by Natoli et al. (2012) are lower than the 92% determined by Mansfield et al. (1999), termination rates still remain high.

There are also many families, as implicated by Skotko earlier, who receive the news following the birth of their child. Skotko used the same survey as mentioned earlier in this review to examine postnatal care. His article "Mothers of Children with Down Syndrome Reflect on Their Postnatal Support" discusses healthcare providers' practices and their training regarding the topic of Down syndrome. From the survey, "mothers reported that their physicians talked

little about the positive aspects of DS and rarely provided enough up-to-date printed materials or telephone numbers of other parents with children with DS" (Skotko, 2005b, p.64). In an effort to determine how parents could receive better postnatal support, Skotko, Capone, and Kishnani (2009) reviewed various articles and surveys that reflected parents' wishes for their postnatal care. In their review, Skotko et al. (2009, p. e754) found that parents desired "access to complete and accurate information for the following questions: What is DS? What is its cause? And, what does it mean for a family to have a member with DS, in practical terms?". Parents also wanted that information to be "balanced, realistic, and contemporary given the current possibilities for people with DS in today's society" (Skotko et al., 2009, p. e754).

Many expressed their frustrations at being presented with only the negative aspects of the condition and "resented when the information was delivered in a manner perceived to be insensitive, unkind, or unconcerned with the welfare of the mother" (Skotko et al., 2009, p. e754). Language and word choice is also vital when presenting a diagnosis of Down syndrome. Skotko et al. (2009) found that many parents felt the following about how the diagnosis was delivered:

> The use of language conveying pity (eg, "I am sorry to have to tell you this, but…"), personal tragedy (eg, "Unfortunately, I have some bad news to share…"), or extreme sorry (eg, "I know this might seem like a devastating loss…") was considered unnecessary and not always reflective of mothers' emotional states. (p. e754-e755)

Instead, parents preferred that physicians offered congratulations as their first words. Another wish for the first conversation was not to be presented with "too much information about possible medical conditions occurring later in life, such as obesity…and Alzheimer disease," which could be too overwhelming initially (Skotko et al., 2009, p. e754). The first conversation

is filled with enough information for families, most of it entirely new to the parents. It is recommended that a follow-up appointment be set up a few weeks following the diagnosis to go over more information as well as provide parents time to ask questions they have come up with since receiving the news (Skotko et al., 2009). It is also best if physicians do not compare a newborn with a diagnosis of Down syndrome to a newborn with a different condition. Whether the diagnosis is delivered prenatally or postnatally, "parents are not typically interested in how their situation is better than or different from having a child with another type of disability" (Skotko, Levine, & Goldstein, 2011, p. 2336).

Because of the importance of delivering a diagnosis in the right manner, Dent and Carey (2006) collected parents' suggestions for physicians to follow when delivering a difficult diagnosis and reported:

> The parents' overall preferences include the following: (a) wishing for more communication of information, (b) for the display of feeling from their physician, (c) more time allowed for parents to discuss the information and ask questions, (d) more expression of confidence in delivery, and (e) parent-to-parent referral (preferred by 87% of the families). (p. 175)

All of these recommendations from families can help ensure parents receive better postnatal care after discovering their child has Down syndrome.

Skotko (2005b, p. 65) also surveyed physicians and discovered that "many clinicians admit that they have little, if any, training on how to deliver such information in a sensitive manner". Because of the need for training requested by both parents and physicians, various training programs are being developed. One such program is a Web-based tutorial that is designed for "improving pediatric residents' skills and comfort with delivering an unexpected

diagnosis of DS," encompassing both a prenatal and postnatal diagnosis (Lunney, Kleinert, Ferguson, & Campbell, 2012, p. 385). After distributing the tutorial to ten pediatric residency training programs around the country, participants "demonstrated a significant improvement in knowledge as a result of using the tutorial" (Lunney et al., 2012, p. 387). When examining the results, it was also discovered that the first-year residents gained the most knowledge from the tutorial (Kleinert, Lunney, Campbell, & Fergusson II, 2009). Based upon these results, Kleinert et al. (2009, p. 328.e5) commented that potentially the "best time to introduce this material is early within residents' training". There are also other more extensive tutorials, such as CD-ROMs and other Web-based tutorials, which provide participants with interactive training simulations. These tools can be helpful in not only creating experiences for residents but also improving confidence levels before actually delivering the diagnosis for the first time. However, it is also best for physicians to receive continued education as diagnoses and information changes over time.

Support for Families

Receiving the diagnosis of Down syndrome is very difficult for parents, even if that diagnosis is given prenatally. Parents envision the perfect child, and any deviation from that image blindsides them. With 1 in every 691 babies being diagnosed with Down syndrome, many parents find themselves asking how this could happen to their baby when 690 other infants managed to "escape" the diagnosis. Depending on the parents' knowledge of the disorder and the support systems offered to them, they can feel like the only people in the world tackling the infinite unknowns that plague the short phrase: "your child has Down syndrome". What is Down syndrome? Is there a cure? How will this affect our child's future and our family's future

together? What are the health problems associated with the disorder? Will my son be able to play catch as I imagined? Will my daughter host imaginary tea parties? Will our child know how to ride a bike, swim, jump, read, and write? The list of questions is endless, and following a diagnosis of Down syndrome some of them can be answered, such as those regarding the condition and immediate health concerns. However, many will have to be answered in time after the child reaches developmental milestones.

With such a large number of questions going unanswered, parents need to find some sort of anchor and know that they are not alone. Their child was the one in 691 infants to receive the diagnosis. That means that 690 infants do not share that same characteristic as their child, but in the next 691 children, one infant will also have Down syndrome. And in the next 691, there will be one more infant with the diagnosis. This trend continues on, and for each infant there are parents and family members attempting to absorb a similar situation with similar questions and fears. To some, these numbers may not be too reassuring. However, when looking at the number of babies born each year in the United States, there are nearly 6,000 children born with Down syndrome based on the statistics (National Down Syndrome Society, 2012). That is in the United States alone and only in one year. Currently, there are around 400,000 Americans who have Down syndrome (National Down Syndrome Society, 2012). Of those 400,000, there are individuals of all races, religious beliefs, socioeconomic backgrounds, genders, and abilities.

At first, it may seem to parents that they are all alone. The decisions they are facing and the questions that prevent sleeping and eating are solely their burdens. While it is true that each child is unique in some way and every situation is different even in the slightest degree, there is at least a handful of individuals in those parents' areas that can provide some insight and some support during this overwhelming time. Support groups are scattered throughout the country to

help provide families with the resources they need to make the initial decisions following a diagnosis of Down syndrome and support beyond those first few months. Hospitals and health clinics can help provide families with this information immediately following a diagnosis of Down syndrome. Also, various groups dedicated to raising awareness and providing support for families can be found in most states.

In a report by the United States Government Accountability Office, it was discovered that parents are more likely to receive resources regarding health and medical problems versus resources that are centered around the family's understanding of the disorder following a diagnosis of Down syndrome (Bascetta, 2010). Parents should receive balanced materials that focus on the different aspect of the diagnosis, not simply the health risks. If there is a lack of balanced materials, some advocacy groups reported that they sometimes "provided the missing material" to local hospitals in the form of "New Parent Packets" (Bascetta, 2010, p.26). It is also important for families to receive information about Down syndrome beyond the diagnosis. The report mentions that "families were likely to receive only about one-quarter (6 of 23)" of the recommended resources for early childhood by advocacy groups (Bascetta, 2010, p. 26). If health clinics do not provide this information beyond the initial diagnosis, it may be difficult for parents to receive this information.

The National Down Syndrome Society, as well as several other national advocate groups for individuals with Down syndrome, strongly encourages families to contact other families with children with Down syndrome. This provides parents with that connection they need in order to answer many of those unknowns that began to arise following the discovery that their child will have or has Down syndrome. Information given to parents at the hospital or health clinic can also provide parents with ways to get in contact with other individuals experiencing a similar

situation. Doctors and nurses can tell parents only so much; unless he or she has a child with Down syndrome, then there is no way they can possibly know how it feels to raise a child who has Down syndrome. It is important for new parents to make that connection with parents who can answer their questions as well as provide insight on questions and experiences that new parents have not even thought of yet.

There are also several printed materials, videos, and webinars available to families in order for them to obtain more information beyond what is presented at the time of the diagnosis. Materials range in information on how to raise a child with Down syndrome to sibling relationships to taking care of one's self as a parent. These resources are wonderful tools for families and provide helpful information and advice. Pairing these resources with connections with other families with children who have Down syndrome enables parents to receive support from a variety of sources. Different support groups also facilitate various activities for families that they can participate in together. Activities depend on the families involved and what they want to set up and participate in, such as a movie night, outdoor games, a parent social, and so forth. The report from the United States Government Accountability Office also mentioned how different groups "provide social development opportunities for children with Down syndrome by hosting playgroups, providing information about the Special Olympics to families, and sponsoring members to attend national and state conferences" (Bascetta, 2010, p. 30). Baseball leagues and swimming classes are more examples of social opportunities available specifically for children with Down syndrome (Bascetta, 2010). The possibilities are endless and are tailored to each group. Combining these various resources help to relieve that overwhelming feeling most parents experience after receiving a diagnosis of Down syndrome.

There are some limitations, however, to the support available to families. In the report by the United States Government Accountability Office, there are some "barriers faced by families related to inaccurate information, financial issues, language, and transportation" (Bascetta, 2010, p. 32). Many of the advocacy groups that reported these issues were taking measures to eliminate these barriers, such as providing educational outreach programs to health care professionals, financial advising workshops, translated materials, and satellite community groups (Bascetta, 2010). If more advocacy groups around the country can make similar improvements, many of the barriers faced by families who need better support could be eliminated.

Discussion

As illustrated by the literature, receiving a diagnosis of Down syndrome can be overwhelming and terrifying for parents. No parent wishes for their child to have a diagnosis when he or she is born. Again, perfection clouds all ideas surrounding the arrival of their child and future years they will share together. A diagnosis of Down syndrome seems to ruin these dreams. For several parents, termination of the pregnancy appears to be the best option for their families. This may be true; however, sometimes their judgment is clouded because they do not have all of the facts or are rushed into making a decision. If a parent is led to believe that his or her child were going to be a "vegetable" with no concept of love or family, and the child would not really get much out of life, then the parent may feel that termination is the best option. However, this future is not going to occur. Children with Down syndrome are not "vegetables;" they do not simply exist and receive nothing from the world around them. How the diagnosis is delivered is vital, and presenting accurate and current information to families enables parents to make more informed decisions when it comes to the future of their child. As Skotko et al. (2011,

p. 2335) mentions, "sometimes, a couple's decision to continue or terminate a pregnancy hinges on the information provided by their health care providers—however accurate or inaccurate, complete or incomplete, up-to-date or outdated". The decision to terminate the pregnancy may also be reached because families have little time to discuss their options. If parents receive the diagnosis further along in the pregnancy, they are on a strict timeline to decide whether or not to abort the fetus before that window of opportunity closes.

As mentioned earlier, Down syndrome is a chromosomal condition that impacts a person in several different ways. Physical appearance, health, cognitive abilities, and other aspects are affected. However, probably the most important aspect of the diagnosis is that it is simply that: a diagnosis. Down syndrome is not the person's identity; instead, it is a part of that individual. It is not realistic nor is it expected to try and forget that someone has Down syndrome. But it is important to put the individual first before his or her diagnosis and remember that he is a kindergartener who loves to play soccer and watch cartoons or that she is a teenager who loves to change outfits three times a day and listens to her music a little too loudly. Every individual is unique in some way, and people with Down syndrome simply have many characteristics that tend to stand out. Physicians need to keep this in mind when delivering a diagnosis of Down syndrome. It is often difficult in a medical setting to focus on other aspects other than the complications of the disorder or how far behind the infant will be in reaching developmental milestones. However, health care professionals must look beyond these very important aspects and include the abilities as well as positive aspects of an individual's life with Down syndrome in addition to the health concerns.

Receiving a diagnosis of Down syndrome does not mean that parents' dreams for their children must come to an end. Their son may still want to play catch, and their daughter may still

want to host imaginary tea parties. A diagnosis of Down syndrome does not threaten these dreams but instead sends families down a slightly different path. Emily Perl Kingsley wrote an essay to help others try and understand raising a child with a disability. Kingsley's son has Down syndrome and in 1987 she wrote a piece entitled "Welcome to Holland," which is often provided to parents of children with disabilities. In her essay, Kingsley equates raising a child with a disability to planning a trip to Italy and ending up in Holland. She explains that the arrival in Holland was not planned, and "you must go out and buy new guide books...[and] learn a whole new language" (Kingsley, 1987). After looking around Holland, there is the realization that Holland has windmills and tulips and Rembrandts (Kingsley, 1987). But yet "everyone you know is busy coming and going from Italy," (Kingsley, 1987). Kingsley ends her essay by advising readers that "if you spend your life mourning the fact that you did [not] get to Italy, you may never be free to enjoy the very special, the very lovely things...about Holland" (1987). No one plans on having a child with a disability or more specifically Down syndrome, but life does not always go as planned. While a diagnosis of Down syndrome may come with hardships, such as medical concerns or delays in typical development, it is not a tragedy.

To ensure that parents do not feel that Down syndrome is a tragedy, it is recommended that parents be provided with complete and accurate information about the diagnosis. This means presenting an unbiased diagnosis to parents, as well as providing them with recent materials to help parents decide what is next for their families. Doctors also need to remain up to date on their information as well; if they do not, then they risk providing false information to families. People carry misguided beliefs that doctors know what is best and will not lead them wrong. Unfortunately, if professionals do not receive the initial training or continued education to remain informed on current information, then physicians will pass along inaccurate information.

Training for residents and continued education throughout physicians' careers can help to eliminate this. These recommendations can lead to fewer parents and professionals believing that a diagnosis of Down syndrome is a tragedy.

Conclusion

Down syndrome may be the most common genetic condition; however, more awareness needs to be raised. First, knowing what Down syndrome is can provide individuals with much more insight, rather than simply fearing the unknown. Also, new parents need to be informed in an unbiased way of the different assessments that diagnose Down syndrome in order to choose which way is best for their family. Following a diagnosis of Down syndrome, parents must also be presented options in an unbiased way. This includes the options given to parents following the diagnosis, such as termination of the pregnancy, adoption, and keeping the newborn. If Down syndrome is explained to parents by only listing off the disabilities the child will have, whether those are correct or not, parents are already seeing their child or at least their child's disorder as a fearful and potentially tragic diagnosis. Support must also be given to every family following a diagnosis of Down syndrome. No parent should feel alone in the process, and he or she should be given as much information as possible in order to prepare for the future. Even though Down syndrome may appear to many individuals to be a tragedy, with more education for the general population and more training for health professionals, this misconception could be eliminated.

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