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English 100
November 18th, 2024

The Power of Words: Transforming the Narrative of a Down syndrome Diagnosis

Every year, thousands of parents across the world receive the life-changing news that their child has Down syndrome—news that is often delivered with little compassion and even less hope. Down syndrome is a chromosomal anomaly in which there is an extra full or partial copy of chromosome 21. Some characteristics of Down syndrome include an infectious smile, powerful, soul-touching hugs, and an impressive ability to overcome challenges. The birth rate for children with Down syndrome has decreased from 1 in 691 (Sheets et al. 434) to as low as 1 in 1,100 (Statistics Canada), largely due to increased prenatal screening (Huete-García and Otaola-Barranquero 8). While the prevalence of Down syndrome has changed, the manner in which the diagnosis is communicated to parents remains a critical issue. A close friend has a sweet little girl who happens to have Down syndrome, and I have witnessed firsthand the joy she brings to everyone around her. However, when she was 25 weeks pregnant, she received the news of her daughter's diagnosis while driving on the highway, in a way that was devoid of hope and included a reminder that it was not too late to terminate. Unfortunately, this experience is far from unique. My proposed solution is three-fold: hope must be integrated into the conversation, accurate resources must be provided, and the news must be delivered by a trained and empathetic professional. By incorporating these strategies, parents will feel better equipped to navigate the challenges ahead, armed with accurate information, emotional support, and connections to the larger community of families who have children with Down syndrome. Ultimately, the way in which parents receive the news about their child's Down syndrome diagnosis often leaves them feeling discouraged and uninformed, highlighting the urgent need for more empathetic and informative communication strategies from healthcare providers.

The emotional distress experienced by parents upon receiving a Down syndrome diagnosis exposes a critical flaw in how healthcare professionals communicate this news, emphasizing the urgent need for more compassionate and supportive information-sharing practices. The way difficult news is communicated can profoundly influence a parent's emotional response, their chronic stress levels, and even their relationships with both their child and partner (Mugweni, 507). Additionally, Ijaz states that "a major concern of the Down syndrome/disability community over the last 30 years is that for every positive diagnosis experience, there are three negative experiences" (2).

As medical technology advances, early, non-invasive prenatal testing for trisomy 21 (Down syndrome) is becoming more common. However, the growing availability of these tests raises serious concerns about how the results are communicated. Research has shown that, after receiving a positive test result, 86% of genetic counselors mentioned termination as an option while only 37% suggested continuing the pregnancy (Ijaz et al. 2). Some families have reported feeling pressured to terminate their pregnancy, describing it as a traumatic experience when healthcare providers repeatedly suggested abortion. In fact, one family shared that they were offered an abortion fifteen separate times after receiving a prenatal Down syndrome diagnosis (Jones). This raises an important question: why is Down syndrome treated as a terminable diagnosis? Sadly, we are at a point where the option to terminate a pregnancy is offered up until the moment of birth (Jones). This trend is seriously concerning, as it signals the normalization of termination as a response to a Down syndrome diagnosis. For this reason, this conversation is deeply important because all lives are valuable, and healthcare professionals take an oath to do no harm, respect life, and support families (Parmar and Rathod 11). To change these outcomes and the overall narrative, healthcare providers must be trained to deliver this news in a manner that is both compassionate and supportive. Every child with Down syndrome deserves to be celebrated, and healthcare professionals must play a key role in ensuring that parents receive the information in a way that instills hope into the conversation.

The lack of information about resources and support networks for children with Down syndrome exposes a significant gap in communication provided by healthcare providers, highlighting the need for more comprehensive guidance for families. Despite the release of the *Practice Guidelines for Communicating a Prenatal or Postnatal Diagnosis of Down Syndrome* (2011) over ten years ago, a follow-up study found that only 12% of families were given a handout or website to explore more information about Down syndrome, and only 22% were informed about available government programs (Ijaz et al. 2). This lack of support is deeply troubling, especially since some families reported that genetic counselors promised to send them information about support groups, yet they never received any follow-up materials (Ijaz et al. 6).

The good news is that there is no need to reinvent the wheel in providing families with the resources and connections. Numerous organizations already exist that offer publications, websites, and phone lines specifically designed to connect families with support and guidance. Healthcare providers could easily direct families to existing resources, ensuring that they are well-informed about available support networks and that they are connected with another family who has an amazing child with Down syndrome. A friend of mine shared how impactful it was to be put into a virtual “birth group” with other families from all over the world who had a baby with Down syndrome in the same birth year as her daughter. It was a huge source of hope and support, and it ensured that she did not feel alone on her journey. Ultimately, all these considerations—access to information, connecting families with peer support, and providing follow-up materials—will make a significant difference for families when they receive the news of their child's diagnosis.

Down syndrome diagnoses are frequently delivered in ways that are insensitive and lack a family-centered approach, leaving families to navigate the emotional impact without adequate support. Research shows that healthcare providers have not consistently implemented a patient-centered approach when delivering prenatal diagnoses. In fact, experts recommend that, before delivering the diagnosis, parents should be engaged in a conversation about how they want the news to be communicated—where, when, and with whom. This personalized approach allows healthcare providers to create an individualized plan tailored to each family's unique needs, ensuring that they feel safe, cared for, and well-informed (Douglas et al. 866).

Furthermore, it is crucial to recognize that fathers are playing a bigger role than ever, in families raising children with Down syndrome. Historically, fathers have been excluded from these conversations, including when the diagnosis is delivered. However, today, fathers are asking to be included, respected, and treated as an equal part of the family equation. As one study noted, dads want to be acknowledged by healthcare providers and involved in every step of the process, especially when it comes to the delivery of such important news (Docherty and Dimond 670).

Additionally, we need to address the method of delivery. A study conducted by Douglas and others provided some results that I found surprising, ten out of fifteen mothers, when compared to oncology patients in similar studies, preferred receiving the news via telephone rather than in person (862). While the emotions experienced in both settings were similar, mothers cited the convenience, ability to process the information in private, and the less intense emotional atmosphere of a phone call as reasons for their preference. (Douglas et al. 863) The takeaway here is clear: the method and setting for delivering the diagnosis should be personalized, not a one-size-fits-all approach. Instead, it needs to be individualized according to the family's preferences and needs. Ultimately, families deserve autonomy in how their story is written, who is involved in the conversation, and where and when it takes place. While this might require additional time and effort from healthcare providers, it is a basic ethical responsibility to ensure that families are treated with respect, dignity, and care. This is just the beginning of the ongoing conversations about their child's health and future—so it is essential that the delivery of this crucial news is presented with the utmost sensitivity and respect for each family's unique situation.

The way a Down syndrome diagnosis is communicated affects every family, whether received prenatally or at birth. Every family deserves a diagnosis framed with hope, optimism, and excitement for the future. In fact, 99% of people with Down syndrome who were asked if they are happy with their life said "yes" (Stotko et al. 2360), providing a powerful testament to the joy and fulfillment that can come from living with Down syndrome. The solution is clear: trained healthcare providers must deliver news of a Down syndrome diagnosis with empathy and compassion, while focusing on connecting families to resources that will encourage, empower, and inform them about the incredible future ahead. It is time for healthcare systems to prioritize empathy and inclusion, ensuring that all children—regardless of their genetic makeup—are celebrated and embraced from the moment they are conceived. The loss of individuals with Down syndrome from our society would rob us of a unique source of love, perspective, and strength. As some countries move toward eliminating Down syndrome entirely (Klucznik and Slepian 46), we must ensure that we celebrate and embrace people with Down syndrome, rather than viewing them as burdens. I feel blessed to have had the opportunity to love a number of people with Down syndrome, an experience that has profoundly changed my life for the better.

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