

In a Spot to Break that News? Delivering a Diagnosis of Down Syndrome to an Unaware Family

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ABSTRACT

This communication presents the views of a cardiologist mother who is raising a child with Down Syndrome/Trisomy 21. She has described her initial emotional struggles and then the subsequent journey, which has required her best efforts to support the child. In this article, she has focused on how medical professionals can make an impact by interacting with families in a positive, encouraging way, and has listed resources that may be of help. She reminds us that there is considerable variability in the outcomes of these children, and hence, not to unnecessarily begin with a negative view of the prognosis. It is important for us to not give up; society must come together, and courageous efforts can help optimize the care for these children and families.

Keywords: Alpha fetoprotein, Atrial septal defect, AV canal defects, Babies with Down Syndrome, Beta-human chorionic gonadotropin, Delivering the diagnosis of Down Syndrome, Dimeric inhibin A, Down Syndrome Education International, Down Syndrome Federation of India, Fetal echocardiography, John Langdon Down, Level 2 ultrasonography, Mongoloid, National Down Syndrome Society, Patent ductus arteriosus, Penta screen, Prenatal counseling, Trisomy 21, Unconjugated estriol, Ventricular septal defect.

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KEY POINTS

- Counseling a family with a fetus/newborn with Trisomy 21 can be difficult.
- It is important to carefully choose words when informing families about their fetus/infant having Trisomy 21.
- The management of a child with Trisomy 21 becomes easier in most cases after the first 2 years, and so an optimistic outlook is well-justified.
- There are several well-established medical/social resources that can be helpful.

COUNSELING PARENTS ABOUT HAVING A FETUS/NEWBORN INFANT WITH DOWN SYNDROME

Clinicians in newborn units often find themselves in a spot where they have to break the news to a family that their newborn infant may have Down Syndrome. It isn't easy and there isn't a perfect way to do it.

Being a mom to a child with Trisomy 21 and a medical professional at the same time puts me in a relatively unique position from where I can feel the joys and apprehensions, fears and elations of a parent. But I also need to look at the various aspects from a factual medical perspective.

Down, not Down's.¹⁻³ Down Syndrome is named after the English physician John Langdon Down,⁴ who characterized the condition; he himself was not affected by it. An apostrophe with "s" connotes ownership or possession.³ Personally, I prefer the nomenclature Trisomy 21 – it is more neutral and does away with the insinuating implications of the word "syndrome." Why a bias to start with?

As an active member of a Down Syndrome parent support group, I have had the privilege of interacting with many parents

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of kids with Trisomy 21. Having myself traversed this journey, I can feel the desperation of these new parents to hear some hopeful words. At the same time, I can also see my colleague's apprehensions of giving false hopes to the parents about the future of their child.

So when I was asked to write for this esteemed journal, I decided to take this opportunity to bring forth a parent's perspective who has been raising a child with Trisomy 21 for over a decade and who also understands the dilemma of a pediatrician/neonatologist/radiologist/obstetrician/cardiologist who at times during their career are put in a spot to "Break the News" of a child having Trisomy 21 to her/his parents and family.

Hope this helps.

My Own Journey

I will first share my own story...

During my second pregnancy, my Penta screen (α -fetoprotein, β -human chorionic gonadotropin, unconjugated estriol, dimeric inhibin A), fetal echocardiography, and a level 2 ultrasonography missed a ventricular septal defect, an atrial septal defect and all other sono-morphological features of Trisomy 21 itself!



Fig. 1: My son with Trisomy 21 (center, identification card with red borders). He is doing well emotionally, socially, and academically. We should never prematurely give up on our expectations of the future of a child, and therefore, the choice of words for delivering the information of this diagnosis to an unaware family is important. The possibilities are enormous and are continuously improving

In retrospective I would say I was fortunate...

Why? Because all of us know about the likely outcome of this pregnancy had we been informed about the diagnosis of Trisomy 21 prior to birth. Nonetheless, there are instances where parents have decided to give birth to fetuses and even twins with Trisomy 21.

As medical professionals, we are mostly involved with the diagnosis and management of surgical or medical issues. It might only be a brief snapshot of these patients' lives. We might not be able to see how their life shapes up; the beautiful, shared moments that the future holds, and the potential that these infants carry despite all difficulties. The first few years are difficult, but life may not be so bad later. Due to a lack of information and the *almost certain* uncertainty of bringing these kids to life, our algorithm has only one answer to pregnancies with chromosomal abnormalities—medical termination.

And that's precisely why I am writing this post.

When I got the diagnosis of Trisomy 21 in my newborn, it was hard to believe. At the outset, there was the natural non-acceptance – denial – questioning (why me?) – and heart-wrenching devastation, in that order. Mostly because...

I knew very little about what lay ahead. I was given a huge responsibility which I wasn't prepared for. And that was all about it.

I wish I had an instruction manual back then.

But once that initial trauma and shock wore off, things started to fall into place. My son began reading at 2 years of age (way before many of his neurotypical peers), following a mainstream syllabus, learning to skate, and riding a bicycle. He goes to one of the finest mainstream schools, the same educational center that accepted his older "scholar" sister. He is his teachers' pride, a favorite amongst friends and neighbors (Fig. 1). And above all, he is the greatest stress-buster of the house. I can easily say he is the only one in the house who is able to take away everyone's stress, without absorbing any himself!

It would be dishonest to say that raising a child with different needs, is a cakewalk. It never was and it will never be so. But there are rewards. Enormous.

And this is not just my story. Anyone who has happened to be around anyone with Trisomy 21 resonates with what I am saying. In

today's era, people with Trisomy 21 are proving to be good students, sports personnel, professionals, and most importantly, wonderful human beings. Thanks to my son, I have been very fortunate to have met many amazing pediatricians and wonderful people. I sincerely hope and pray that every family blessed with a kid with Trisomy 21 finds comfort and hope in their pediatrician and in the larger society.

My Humble Suggestions

This is what I have learned. My most humble request to my medical colleagues—whosoever lands in a position to deliver a diagnosis of Trisomy 21 to a family:

1. Please always remember that the parent "IS NOT" prepared to hear what you are going to say!
 - a. Please be very sensitive—this is going to change their lives forever.
 - b. Words should be chosen carefully. Using the right words at the outset is important; these will make a lasting impact.
2. Parents must receive the news together.
 - a. Please ensure they are holding the baby when they get the news and address the baby with her/his name if it has been decided.
 - b. "Baby has Trisomy 21" is a better way, than to say that it's a Down Syndrome baby.
 - c. The word "Mongoloid" should never be used. It is also legally prohibited.^{5,6}
 - d. A good way to begin is by congratulating them for their new baby and not with "I am sorry." Consistency is important; the attending pediatrician and obstetrician should convey the same message. Other staff members should preferably leave the discussions to the clinical leaders and not share their personal opinions with the family.
3. Every city across the globe has existing parent support groups for Trisomy 21. It will be a great idea to have an experienced parent on board while informing/counseling the parents. Please try.⁷

At that point of time parents will connect with other parents better than the professionals.⁷

4. Please tell them that it is not a catastrophe to have a baby with Trisomy 21.

It is OK and it is doable.⁸

5. Having a baby with Trisomy 21 has nothing to do with any of the parent's wrongdoing/karma/previous life. THIS IS NATURE. Statistically, every 700–1000th child will have one extra chromosome and this child just happens to be that 1000th child, that's it.⁸
6. This is the most frequently seen and most manageable congenital condition.⁹ This is NOT a disease!
7. There might be health issues related to cardiac/endocrine/hearing and visual function. However, these are more difficult during the first 2 years, and subsequently, the health is more likely to follow a better track.^{9,10}
 - a. In the absence of compelling reasons, the family should be sent home early.¹⁰ They'd need some time together.
 - b. Parents should be encouraged to come for follow-up visits to screen for possible medical concerns.¹⁰
8. Many health issues, such as cardiac lesions can be permanently dealt with, in one go.¹¹ Others are more easily correctable during follow-up visits.



Fig. 2: These two photographs again emphasize that the possibilities are enormous. I personally cannot do many things that he can!

9. Physical milestones and intellectual delays are expected¹² but a lot can be done about these (Fig. 2).
10. Lifespan can be near normal in today's era of ever-improving medical facilities.¹³
11. There is no huge financial liability that comes with this condition; most issues can be handled with "common-sense parenting."¹⁴ Having a child with Trisomy 21 does not automatically mean that there will be a permanent need for the parents to see doctors and therapists all their lives.

Parents are the best, most effective, least expensive, and most easily accessible therapists!

12. "Better qualified and financially sound families living in bigger cities can do a better job raising kids with Trisomy 21" – is a myth.
 - a. Common sense has nothing to do with qualification, wealth, or facilities available in metropolitan areas.
 - b. I would really like to emphasize this. All training protocols work best and are most effective when done at home, by the family, and in the natural environment of the child.

Families with most testing conditions have often shown the best results!

13. Having a child with Trisomy 21 DOES NOT necessarily mean that either/both parents would have to sacrifice their professional growth. Please relax! Having a child with this diagnosis does not automatically imply this; most setbacks are likely to be temporary. The initial years will demand more care and time,¹⁵ a little more than a typical newborn would in catching up with the milestones and language development.
 - a. The special needs gradually improve in most children. If both parents and the extended family decide to work in harmony, it will work wonders for the kid and also enhance the bonding among the family members. They will mostly be able to reorganize their schedules, manage the situation, and realize their professional dreams too. But, for this to happen, we as professionals need to hold hands and remove the negative misconceptions from new parent's minds.

- b. *Soon enough, the parents will know more about Trisomy 21 than professionals! All of us, need to regularly update ourselves, especially before we are to counsel a new family.*¹⁶ Moralistic professionalism.
14. We need not always be too factual in one go. The flow of information can be gradual, or swift, per the needs of the situation.
15. Guide them on what to google. I have found many authentic websites, books, and resources. A few of these are:
 - a. #NDSS (National Down Syndrome society);¹⁷
 - b. #Down Syndrome Education International;¹⁸
 - c. #Down Syndrome Federation of India;¹⁹ and
 - d. #Babies with Down Syndrome "A new parents guide" book/e-book.²⁰
 - e. There are many other books dedicated to each developmental area: gross and fine motor skills, language, nutrition, adolescence, just to name a few. Google can be useful – a googol number of possibilities!
16. We live in an era of ever-improving medical facilities and freely-available, evidence-based knowledge. Thankfully, the internet has democratized the access to information and knowledge. It has the potential to keep parents abreast with the latest global advances in management.
 - a. Individuals with Trisomy 21 have shown improvements in cognitive function and independence over time.²¹
 - b. Appropriate and timely intellectual stimulation helps.

Undoubtedly, it would require hard work, dedication and patience from the family. But medical professionals have bigger shoes to fill as besides providing necessary medical care we need to give them a sympathetic ear and wise counsel, hold their hands in times of distress and motivate them to help the individuals with Trisomy 21 fulfil their destiny. Let's spread hope together and avoid being remembered by the family as a pessimistic clinician.

Please try not to predict the future of the child, for the possibilities of growth are enormous.

NEVER GIVE UP ON ANY CHILD! Every baby counts!

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