



Neonatal Ethics Teaching Program

Scenario Oriented Learning in Ethics (SOLE) Announcing the Diagnosis of Trisomy 21

Facilitator Guide

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Description of SOLE Workshop

Delivering news to patients and their families can lead to emotionally charged conversations that are difficult for trainees to feel comfortable or effective in. Trisomy 21 (T21), or Down syndrome, is the most common viable chromosomal anomaly. Although prenatal screening exists, over 80% of T21 diagnoses continue to be made postnatally to unsuspecting parents who report a desire for better communication from healthcare professionals when they first receive the news of their child's diagnosis.¹⁶

To better equip trainees to thoughtfully and professionally deliver a diagnosis of T21, we have designed a workshop founded on the principles of Scenario Oriented Learning in Ethics (SOLE) which introduces a framework combining current evidence on communicating life altering news with stated parent preferences. During the workshop, trainees will have the opportunity to practice using the provided communication framework with a standardized patient trained as a new mother. By personally working through a clinical scenario as well as observing workshop colleagues doing the same, participants will gain insight into what is effective as well as ways to troubleshoot encountered pitfalls in a safe and supportive environment. By pre-emptively practicing evidence-based communication, we hope to increase trainee comfort with critical conversations and improve parents' feelings regarding the quality of communication and support provided while receiving real life T21 diagnoses.

Objectives

1. Inform families of a T21 diagnosis in manner that is sensitive to their cultural, social, and religious backgrounds.
2. Recognize and validate a variety of emotional reactions even if they are not shared.
3. Utilize current evidence to explain T21 in a practical and tangible manner.
4. Incorporate shared decision making into a focused evidence based management plan

Instructions to Trainees on How to Prepare for this SOLE

1. Read the required readings.
2. Review the evidence on how to improve delivery of T21 diagnosis (see Appendix A)
3. Read, in detail, the case scenario involving the standardized patient (see Appendix B)
4. Review, in detail, the suggested communication strategies outlined in the provided communication framework (see Appendix C).
5. Read the summary sheet entitled "Knowledge Based Principles" that addresses common questions parents may have related to T21 (see Appendix D)

Required Reading

- Bull, M., & the Committee on Genetics. (2011). Clinical Report - Health Supervision for Children with Down syndrome. *Pediatrics*. 128(2), 393-406.
- Koh, H.K., & Rudd, R.E. (2015). The Arc of Health Literacy. *JAMA*, 314(12), 1225 - 1226.
- Skotko, B., Capone, G., & Kishnani, P. (2009). Postnatal Diagnosis of Down syndrome: Synthesis of the Evidence on How Best to Deliver the News. *Pediatrics*. 124, e751.

Additional References (non-mandatory)

- Hobson-Rohrer, W., & Samson-Fang, L. (2013). Down Syndrome. *Pediatrics in Review*. 34, 573-574. DOI: 10.1542/pir.34-12-573
- Hedov, G., Wikblad, K., & Annerén, G. (2002). First information and support provided to parents of children with Down syndrome in Sweden: clinical goals and parental experiences. *Acta Paediatrica (Oslo, Norway: 1992)*, 91(12), 1344-1349.

SOLE Workshop Timeline

Introduction (15 min)

- 1) Pre-briefing between training and supervisor to outline workshop goals
- 2) Answering of questions to clarify the strategies for delivery of a diagnosis of T21 (trainees and supervisors)

Practice with the Standardized Patient (80 min)

- 1) 25 min to cover the initial steps of the medical encounter
- 2) 5 min to cover the closure of the medical encounter
- 3) 10 min of discussion

Conclusion (20 min)

- 1) Constructive feedback from standardized patient to trainees
- 2) Constructive feedback from supervisors
- 3) Debriefing to review the key learning points of the practice sessions

Program Evaluation (5 min)

Instructions for Facilitators - How to Supervise a SOLE

A. INTRODUCTION

The supervisor has to:

- Remind the trainees that the session represents a safe learning environment where mistakes are allowed for learning purposes.
- Clarify any of the trainees' questions/comments about the strategies outlined in the communication framework.
- Explain the specific details about interacting with the SP as outlined below.
- Ask trainees to make note of their comments or questions as they are observing other participants' interactions with the SP.

Overview of role-playing with the standardized patient

The role-playing will happen in multiple parts. The supervisor will give instructions during the Introduction as per the 3 sections below:

1. Preparing for the role-playing:

- Ask one or more trainees to play the role of the doctor.
- If more than one trainee will be involved in the encounter, select one participant to start the interview and have the next one complete or modify the ongoing interview after a time out according to the suggestions made within the group.
- Trainees may rotate more than once during their respective parts if that is felt to be beneficial for the group.

Note: The trainee(s) participating will have the *Trainee Guide* in their hands so they have all necessary information to reasonably understand the context and speak to the SP playing the parent.

2. Process during role-playing:

- The trainees role-playing the doctor will have a total of 20-25 minutes to complete the interview.
- Specify that mistakes are allowed and that to forget some aspects of the communication framework is normal.
- Remind the trainees observing the encounter that only the supervisor has the right to interrupt the interview at any time if s/he sees that the trainee acting in the scenario is stuck or if comments need to be made (i.e. a great teaching point is noted).
- Remind the trainee that s/he also has the right to stop the role-play if s/he feels stuck or uncomfortable.

3. Scenario set-up

- Ask the trainee who will begin in the role of doctor to step out of the room.
- Prepare the hospital scene with the pre-organized material (i.e. bed for mother, the cot for the baby mannequin, a chair etc.).
- Call the SP into the room and introduce her in their acting role only to the observing trainees.
- Call the trainee who will be playing the doctor back into the room and begin the scenario with the SP.

B. PRACTICE WITH THE STANDARDIZED PATIENT**During role-playing, the supervisor has to:**

- Keep the workshop on time
- Observe the performance of the trainee
- Interrupt the interaction with the SP as required (see below)
- Maximize interaction time with the SP (i.e. keep debriefing succinct)

When the scenario is interrupted, the supervisor has to:

- Ask the SP to leave the room.
- Proceed with debriefing the trainee who has played the doctor role by asking him/her what part(s) of the experience were easiest, followed by those that were most difficult
Ex. “Can you identify one thing you did well?” and “please, tell me, one thing that you would like to improve next time.”
- Clarify the difficulties or conflicts encountered and reinforce strengths.
- Generate a round table discussion by asking some of the trainees who observed the interview to comment on one specific positive aspect and one aspect to improve.
- Reformulate the comments that were not clear enough.
- Ask the trainee who has played the role of the doctor to summarize at least one of the positive comments and one of the aspects to improve.
- At the end, generate 2-3 options that the trainee can try for the next part of the interview in order to help resolve the difficulties or conflict.

After the debriefing, the supervisor has to:

- Coach the trainee through the next part of the scenario.
- Clarify with the trainee if they are comfortable applying the options.
- Identify the moment of the interview where the SP has to replay the consultation.
- Direct the SP outside the teaching room to restart the interview and if they need to make modifications to their role-playing.
- Invite the SP to come back in the room and restart the scenario.

C. CONCLUSION

The supervisor has to:

- Ask the SP to introduce themselves in terms of their real identify.
- Ask for the SP's feedback to help the trainees in terms of identifying strengths as well as areas that need improvement.
- Ask the trainees if they have questions for the SP.
- Complete and summarize the workshop by asking all workshop trainees, including those who did not interact with the SP, to:
 - Review what strengths and learning points they remember and plan to take away with them.
 - Ask trainees to complete one electronic self-reflection form in the 24-48 hours after the workshop in order to assist their learning.
 - Remind them also to fill out the electronic self-reflection forms after real life situations.
- Thank the SP and the trainees for their valuable input.

Appendix A

Rationale for the Communication Framework

Specific to T21, reviews of the literature on both a national and international level have shown that parents repeatedly remember negative experiences when hearing a diagnosis of Down Syndrome applied to their child.^{3,7,14,15} When preparing to deliver the news, healthcare professionals should consider who, where, and when it will be delivered. When initiating a conversation about an unexpected T21 diagnosis, it is critical that the news not be framed as “bad” while still acknowledging that it is life altering. The first words set the tone for the entire conversation with many mothers recalling what was initially said over two decades later¹⁴ so it is vital that this be done thoughtfully and positively.

In regards to who should communicate the diagnosis, parents report a strong preference for the news to be delivered by a single empathetic physician who is knowledgeable about T21. The diagnosis should be made when both parents are present. Mothers told alone endorsed feeling burdened by the responsibility of sharing the news with their partner and reported great dissatisfaction when their partner was informed before them.¹⁶

Timing of the news has a significant impact on how parents received a postnatal T21 diagnosis. Given the need for early medical intervention in many babies with T21, as well as the fact that classic phenotypic characteristics often raise early concerns, the majority of parents are told within the first 24 hours of life.⁷ Within this time frame, the overall mean parental satisfaction regarding timing of the news was 56% with the majority of parents, 77%, satisfied when they were told within 3-5 hours of their baby’s birth. Strongly negative experiences have been reported when diagnosis was done at times perceived as both “too soon” and “too late” regardless of the absolute amount of time that was allowed to pass between the delivery of the baby and the breaking of the news.⁷ With this in mind, efforts should be made to balance the mother’s recovery after birth with timely initiation of delivering the news.

Prior to delivering the diagnosis of T21, ensure that you are aware of the major physical characteristics associated with the syndrome and review the screening guidelines for children with Down syndrome so that you may address any concerns parents have. It may also be useful to remind yourself of the typical forms of prenatal screening used for T21 (ex. IPS, first trimester screen, nuchal translucency) as parents may have questions about why the diagnosis was not detected prenatally. Remember that T21 is a common diagnosis that is most often determined by spontaneous mutations so parents of children with T21 have varying levels of baseline knowledge and personal intellectual functioning. It is important to meet families’ current understanding and add to them in a way that is appropriate for their lives.

The setting for the conversation should be private, quiet, and free of unfamiliar personnel to promote an open and safe atmosphere for parents to react without fear of judgment.^{3, 16} In particular, visitors and additional healthcare staff should be asked to leave the room. Parents expressed intense dissatisfaction when the news that their child had T21 was shared in front of hospital roommates, extended family, or friends. If a mother is alone, it may be important to

amend this at the mother's request so that a single important person is present for the conversation to provide support.³ Healthcare professionals should also be prepared to provide extended amounts of time to the encounter and work to limit the number of interruptions (ex. pagers) that occur as parents were more likely to remember the experience of receiving the news as negative if they felt rushed or interrupted.¹⁴

Language used to deliver the diagnosis is of critical importance both in terms of what is said and how it is phrased. The SPIKES format is a well-developed communication guideline developed for "breaking bad news" to adult oncologic patients and their families.¹ Recognizing that Pediatric patients are not simply "little adults", it has been adapted over time to include a more collaborative approach that facilitates family-centered conversations.¹⁸ While the basic SPIKES tenets remain relevant to preparing for a challenging discussion, certain aspects have been proven antiquated in helping families to reach a true understanding of what healthcare professional want to communicate in critical conversations. In essence, there has been a shift to recognize that, when faced with challenging or unexpected news, even the most knowledgeable individual may struggle to grasp the full implications of what is being communicated.⁹ With this in mind, there is growing support for implementation of the "teach back method" into the delivery of life altering news which places the burden on practitioners to use proactive methods to check for comprehension ("Help me to see if I've left anything out").^{4, 9} By validating questions as a normal, and even expected part, of a critical conversation, you create an atmosphere that is more conducive to honest dialogue and release families from feeling guilt or embarrassed if they are still not clear about certain details they were told. Essentially, you recognize that communication is not a linear phenomenon as suggested by the SPIKES method but rather a multi-tiered complexity consisting of what is intended, said, heard, and ultimately understood.

For the first conversation, information should be limited to the medical conditions children with T21 are likely to face in their first year of life.¹⁶ The process of receiving an unexpected diagnosis can be overwhelming enough, so information about possible issues later in life should be deferred. Information should be communicated in a practical, confident, accurate, and positive manner. Parents reported feeling resentful of information they perceived as vague, outdated, or overly pessimistic.¹⁶ Terms such as "mongolism" should never be utilized as they are hurtful and inappropriate. The process of hearing the news can be shocking or even traumatic for some families.⁷ As you share the news with them, be ready for a variety of reactions including but not limited to: shock, anger, devastation, helplessness, and rejection.¹⁷ It is important to empathize with families regardless of their reactions and demonstrate your own honest display of emotion as appropriate as parents felt comforted by this rather than deterred.³

Appendix B

Clinical Scenario and Goals of Encounter

Reason for consultation	You have been called by an Obstetrician from the Labour and Delivery Ward. The Obstetrician states that he has just delivered a baby girl in Room 15 to a healthy 28 year old G2T1P0A1L1 mother named Amy. The baby girl was born 3-4 hours ago and the L&D team has concerns. The RN has commented that the baby appears a “little floppy” and that she has had difficulty latching. The OB staff has voiced to you that he suspects the baby may have Down Syndrome given some of her facial features but he is not sure. No one has raised their suspicions to the parents. You are consulted to assess the baby for a diagnosis of query Down Syndrome and deliver the news to the family if it is correct. As you enter the room, you see Amy holding her baby. She is surrounded by multiple people who introduce themselves as friends who are just on their way out. Amy’s partner is not in the room as he has left to bring their parents home. She seems curious as to why you’re here.
Trainee’s Goals Reminder	1. <i>Setting:</i> • Establish a safe and secure environment in which parent(s) feel free to express themselves. • Work to ensure that parent(s) to receive the news together with distractions and non-essential personnel minimized throughout the discussion. 2. <i>Perception:</i> • Clarify if parent(s) have recognized any features that they are concerned about. • Clearly explain your role as well as the reason you have been asked to see them. 3. <i>Invitation:</i> • Offer parent(s) the opportunity to share their main questions and concerns. • Clarify their baseline understanding of T21 so that you may appropriately build on it. 4. <i>Knowledge:</i> • Describe T21 in practical terms that highlight what features make you suspicious of the diagnosis for their child as well as what is “typical” about the child. • Clarify immediate medical concerns and outline corresponding management. 5. <i>Explore and Empathize:</i> • Normalize parental feelings and validate their reactions in a way that shows you appreciate their experience even if you do not feel the emotion yourself. • Adjust the rate of information delivery to the needs of the family. 6. <i>Strategize and Summarize:</i> • Ensure parent(s) have an understanding for the immediate steps that will be taken to care for their child and empower them to get actively involved in their child’s care. • Provide parent(s) with time to reflect on the news and schedule follow-up meetings with as appropriate.

Appendix C

Case Scenario for Standardized Patient

Clothing and setting for role-playing

Comfortable, loose clothes (e.g. 'sweat suit'); hospital gown (provided); you will be sitting in a chair and not too far from you will be a cot with a sleeping baby (mannequin) covered by a blanket. Alternatively, you may be sitting in a chair with the baby in your arms.

Description of standardize patient role

You, Amy, are a 28-year-old woman from Canada. This is your second pregnancy. You have had one miscarriage previously but the current pregnancy has gone smoothly and you are very excited about the birth of your first child.

You regularly saw your family physician here in Ottawa and feel you have a good rapport. You were offered routine prenatal testing but declined it as you had no specific concerns as your family history does not have any specific medical or genetic conditions that you know about. You had a brief period of spotting early in your first trimester and it never recurred. You have no history of hypertension or known diabetes. You don't have any history of health problems. You didn't smoke or take any medication, alcohol, or drugs during this pregnancy.

The fetal ultrasounds done in the first trimester revealed a normal nuchal translucency measurement. The ultrasounds done at the beginning of the second trimester were similarly reported as normal without any notable malformations. Your GBS status was unknown but all your serological results (e.g. Hepatitis B, HIV, etc. ...) so far are negative.

You have been married for two years and your husband, Matt, has been supportive of your pregnancy. There has been some social stress as he works for the Canadian government and frequently travels for work. This will also be his first child and he is very excited about the event. He is otherwise healthy with no contributory medical or surgical history. He was adopted at birth and is not certain of his biological parents' medical history.

You came to the hospital last night because you had contractions and your water broke. You presented at 39+4 weeks gestation in active labor and, after 20 hours of contractions, you delivered vaginally without complications. You hear your baby cry and are overwhelmed with joy! The Obstetrician congratulates you and tells you that you have a new baby girl. After briefly holding her, the nurse brings your baby to a nearby isolette for routine care. You notice the nurse speaking in a hushed tone to the doctor who speaks quietly in return. You feel confused and worried about what is going and ask how your baby is doing. The nurse says she is doing well. Your daughter is bundled in her blanket and brought to you. You and your husband decided early on that if you had a girl you would name her Jasmine and you are pleased that it seems to suit her. You think she is beautiful but notice that she

doesn't quite look like you or your husband. She keeps slipping down in your arms and you have to ask for help with readjusting your hold. You wonder why the doctor and nurse were whispering to one another but block out the worry because you are so excited to finally meet your baby girl.

After you're all settled, your husband excuses himself with your visiting family to drive them home. A new doctor enters the room after they've left and introduces him/herself as the Pediatrician at the hospital. You suddenly feel anxious and wonder what they are here to talk about. You sense that something is about to happen.

Information to help with role-playing

- The doctor will deliver for the first time to you – the mother - the unexpected news of a Trisomy 21 (Down syndrome) diagnosis.
- There are different ways the doctor could interact with you and the baby while beginning the encounter but all approaches should include the following initial components:
 - The doctor should introduce him/herself and encourage unknown or additional people (i.e. RN, acquaintances) to leave the room.
 - They should inquire about your husband and clarify where he is as well as when he will be back.
 - The doctor should ask whether you would like to hold the baby during your conversation together.
 - If you decline, the doctor should place the baby in the small bed and put it close to you so you can still see her face.
 - The doctor should periodically look at the baby while speaking.
- To open the encounter, the doctor should speak positively about your baby and may congratulate you on the birth of your daughter.
 - If words of a particularly negative connotation are used at any point during the encounter to describe your daughter or the diagnosis this should be pointed out during the feedback period.
- The doctor should ask whether you have noticed anything “unexpected” about your baby and may use this as a tool to transition towards delivering the news.
 - You should respond to this by saying that you have found that your baby seems to have difficulty staying in one position in your arms and that she does not really look like your or your husband.
 - The doctor should validate these concerns and comment on other differences noted in general terms with specific elaboration on any particular features you inquire about.
- Prior to delivering the diagnosis of Trisomy 21, the doctor should provide a “warning sign” that alerts you to the fact that they may break some unexpected news.

- Appropriate phrasing may include pointing out specific features in your baby that are common to Trisomy 21 or the use of terms such as “difficult” or “unexpected”.
 - The news should not be framed as “bad” and if it is this should be communicated during the feedback period.
- The doctor should specify that they are concerned that your baby has Trisomy 21 and ask what you know about this diagnosis.
 - An appropriate response is that you have worked with children with Down syndrome as well, as special learning needs as a teacher, but that none of your relatives or friends have been diagnosed with Trisomy 21 nor have they ever had a child with it.
 - The doctor should identify specific physical features of Trisomy 21 that they see in your baby and define the cause of the syndrome using simple and straightforward terms.
 - The doctor should explain the risk factors for Trisomy 21 and, if prompted, clarify why the diagnosis was not detected prenatally.
 - The doctor should not place any undue blame on the family physician even if prompted by you about whether your “family doctor did something wrong?”
 - As you listen to the doctor’s concern, you may continue through the encounter using one of the suggested attitudes outlined below.

1st SUGGESTED ATTITUDE: Anxious and Expressive

- As the doctor starts to deliver the news, you may begin to become **highly anxious**
- You may express your worries through a series of questions and ask about why this happened to you, why all of the ultrasounds during the pregnancy were normal, etc.
- As the trainee attempts to answer your questions, you may continue to interrupt him/her new and additional questions without waiting to hear the appropriate answers
- As your anxiety intensifies, this may be playacted with crying, yelling, or shutting down

Note: We may need to stop the simulation for a 3-5 minute discussion if the trainee struggles with this; you will be asked to wait outside the room.

2nd SUGGESTED ATTITUDE: Cooperative

- If the doctor has demonstrated strong initial communication skills (i.e. clear introduction of their role, positive opening statement, established quiet and supportive environment), you may choose to remain **receptive and cooperative** to the discussion
- At this point, you may start to **focus on your daughter** and cry silently while you hug and kiss her several times (acceptance/bonding is happening) without seeming overly upset or distressed by the news
- Alternatively, you may simply remain pleasant and attentive with open body language and no strongly expressed worries or negative emotions

3rd SUGGESTED ATTITUDE: Shut Down and Detached

- As part of this attitude, you should appear **“shut down”** and maintain a **cold demeanor** both toward the trainee and your baby
- You should use **non-verbal** communication (i.e. avoid looking at your baby, avoid face-to-face interaction with the doctor, avoid holding your baby anymore, show indifference even in your answers) to demonstrate that you are no longer cooperative to the interview
- If you continue to answer questions, you should use short phrases that are difficult to elaborate on such as: “yeah, sure”, “if you think so...”, or “okay”
- The doctor will soon realize that s/he is stuck in a situation with potential conflict, and that s/he will not be able to have your full attention/cooperation any longer

Note: We may need to stop the simulation for a 3-5 minute discussion if the trainee struggles with this; you will be asked to wait outside the room.

4th SUGGESTED ATTITUDE: Confrontational

- As part of this attitude, you may become very **upset and aggravated** by the news and openly express your frustration with rhetorical questions (i.e. **“why did this happen to me?”**)
- You may begin to **raise your voice** and state that you don't wish to proceed with the conversation until your spouse returns or comment that the doctor doesn't seem to care about your particular circumstance
- Alternatively, you may become more **antagonistic** and begin to ask more assertive questions such as **“are you going to take my baby away from me?”** or **“are you saying that my baby is going to die?”**

Note: We may need to stop the simulation for a 3-5 minute discussion if the trainee struggles with this; you will be asked to wait outside the room.

- Regardless of the attitude expressed, the doctor should wait in the room and give you time to express your surprise/emotion/crying/affection etc.
 - If you are visibly upset, the doctor may acknowledge your distress (ex. “I can understand if this comes as a shock”).
 - The doctor may also choose to silently provide you with tissues or offer support if appropriate (ex. placing their hand on your hand).
 - If you feel hurried or pressured to continue the conversation, this should be communicated during feedback.
- As you react to the news, the trainee should display the following communication skills:
 - Allows you time to ‘take in’ the information which may include short periods of silence if appropriate.
 - Acknowledges your reactions and validates your emotions which could range from plain denial and shock to taking responsibility for what happened (ex. “it’s natural to be overwhelmed”).
 - Inquires as to what you would like to discuss first and shares information in a way that is tangible.
- The doctor should routinely pause to ask if there is anything they can help to clarify for you and invite any further questions you have thought of.
 - If you feel that something was not clearly explained, you may redirect the conversation to address it again.
- After explaining the diagnosis, the doctor should transition toward a discussion about what care will be provided for your baby.

- Again the doctor should clearly explain what kind of investigations s/he is planning (ex. blood work, echocardiogram, etc.) and offer a clear timeline for what will be done over the next few days.
- In order to help promote shared decision making in regards to the care plan, the doctor should be open to your position and show respect for what you think and want for your baby.
 - If at any point the doctor has an attitude that is too paternalistic or s/he does not explain in simple terms why the investigations are necessary, this should be communicated during the feedback period.
- If you express that you wish to wait for your husband because you will need some time to speak with him **alone** before any investigations are done, the doctor should acknowledge your desire and:
 - S/he should try to explain that certain investigations need to be done in **the best interests** of your baby and you should expect the doctor to explain what that means and if there is an urgent matter to investigate.
 - Based on this information, you may reconsider your decision and consider going ahead with the investigations.
 - The doctor may also decide to respect your wishes for no investigations at this time or go ahead only with some investigations instead of all of them (i.e. the doctor is trying to compromise and support your wishes).
 - S/he might offer support by asking: “Would you like for me to be present when you will talk about Jasmine’s condition to your husband?”
- The encounter should conclude with a summary of the plan as well as an effort to establish a follow-up time later that day or in the next few days for further discussion.
- The doctor may encourage you to write down your questions and offer to return when your husband is back to review the entire conversation with both of you.

Appendix D

Key Components for the Communication of a T21 Diagnosis

**Note: this is a guideline of steps to facilitate communication. They do not necessarily need to be performed in the order provided. Many steps may occur or re-occur throughout the whole encounter.*

<i>Suggested Communication Strategies</i>	<i>Rationale</i>
* Greet the parents and introduce yourself * Inquire about father's/partner's presence or absence (if applicable)	Clarify your role to establish trustful relationship. Work to inform parents together as this promoted a greater satisfaction with the overall process.
* Choose a setting that is quiet to minimize disruptions (remember to turn your phone and/or pager to vibrate) * Encourage unknown or additional people to leave the room (i.e. RN, acquaintances)	Promotes safe and therapeutic space for what may be a challenging or sensitive conversation. Allows parents to freely express themselves without feeling rushed, interrupted, or judged by others.
* Be sure that the parent(s) have seen their baby and that s/he is in the room with you * Refer to the baby by his/her name and look at him/her as appropriate	Destigmatize the diagnosis as early as possible to promote attachment. Parents should see you role modelling natural interactions with them as well as with the baby.
* Sit down so that you are on the same level as the parents throughout the discussion * Make eye-contact with the parents and maintain a calm but confident demeanor	Reinforces that you are equals and minimize feelings of paternalistic communication. Promotes a therapeutic alliance and decreases the anxiety that can be bred by uncertainty.
* After congratulating parents on the birth of their baby, if you have not already done so, you should deliver a "warning sign" 1. "I've been asked to see you because the OB team has some concerns about your baby's [ex. overall lower muscular tone]..." 2. "While examining [name of baby/your baby], I noticed some features that make me think of Down syndrome [ex. flat nasal bridge, upslanting eyes]..." * Clarify whether parents have noticed anything "unusual" or "unexpected" about their baby and, if so, what they observed	Recognizing that the delicate nature of this conversation may trigger a variety of emotions, parents should begin by receiving congratulations on the birth of their baby as this prepares them to receive unexpected news in an optimistic manner. Mothers frequently report noticing something "different" about their baby even before the diagnosis of T21 is given. Clarify for identified features whether they are typical to T21 or unrelated.
* Clarify how parents understand T21 so that you may build off their existing knowledge base (pay attention to their language) 1. "What have you heard about Down syndrome?" 2. "Have you ever had any personal experience with someone who has Down syndrome?" * Pay attention to the language they use so that you may match the complexity of your explanations to their own understanding	T21 is a relatively common diagnosis so it is important to appreciate parents' baseline knowledge. Studies have shown that only 1/2 of parents felt information conveyed was easy to understand.

<i>* Ask parents what they would like to hear about first and address this issue if they have a preference so that you may focus on answering their concerns and questions</i> 1. <i>“Is there anything you would like to discuss?”</i> 2. <i>“What are your questions?”</i> 3. <i>“Is there anything you would like to understand better”</i>	Allows parents to guide and lead present encounter so that you may address their chief concerns early on. Assume that what parents raise is the most important for them to know at first. Other concerns that are less important for them will come later.
<i>* Be honest but sensitive when disclosing that you suspect their child may have T21 and provide specific rationales as to why</i> <i>Ex. “Some of the physical features [insert specific features] we see in your baby are commonly seen in babies affected by Down syndrome or Trisomy 21”</i> <i>* Define T21 using simple and practical terms</i> <i>Ex. “Down Syndrome is a genetic condition that occurs when someone is born with an extra chromosome”</i>	Mothers report remembering what was initially said over twenty years later. Language should be thoughtful and clear. Be mindful that technical jargon can trigger miscommunication even if inadvertent. Initial explanations should be succinct and easy to assimilate as parents are likely to feel a certain degree of shock that when hearing the news.
<i>* Use a slower pace while delivering the news and observe verbal as well as non-verbal cues</i> <i>*Identify cues using general “gentle” descriptions</i> 1. <i>“I recognize this may be unexpected...”</i> 2. <i>“I can understand if this comes as a shock...”</i> 3. <i>“I appreciate that this news may be startling...”</i> <i>* <u>Avoid</u> offering personal experiences and opinions that are not solicited as these <u>may not</u> reflect how parents actually feel</i>	Parents describe their first reactions to their child’s T21 diagnosis as: shocked, angry, devastated, overwhelmed, stunned, and/or helpless. It is crucial to validate and normalize the initial reactions, which will impact how well the next steps of the encounter will evolve. Be very sensitive to how a child with developmental delay may fit into the varying cultural, racial, social, and religious backgrounds of individual families.
<i>* Acknowledge parents’ emotions and help them to process their own reactions to the news</i> <i>EXPLORE: “How does that make you feel?”</i> <i>VALIDATE: “It’s natural to feel overwhelmed.”</i> <i>EMPATHIZE: “I can see that this is difficult for you... I’m here to help however I can.”</i> <i>* This is <u>not a single step</u> and should be performed as appropriate throughout the conversation</i>	Uses active listening techniques to reinforce the therapeutic alliance and demonstrate you care. Avoid language that apologizes for the diagnosis or implies what parents are feeling (ex. I know this might seem like a devastating loss...). This can seem overly negative and may not reflect how parents truly feel.
<i>* Allow for silence and time without exuding a sense of unease</i> <i>* If you feel the dialogue has broken down or that parents are too overwhelmed to continue, offer to return later</i>	Only ½ of parents report that they felt the physician informing them of their child’s T21 diagnosis gave them enough time to absorb the news.

<i>* After addressing parents questions or concerns, key knowledge points should be communicated to parents as they arise in conversation including:</i> • Individual differences in intellectual functioning and that this cannot be predicted at birth • Specific positive attributes such as warm and caring personalities with an openness to displaying affection • Medical issues in first year of life (ex. congenital heart defects, gastrointestinal atresia, transient myeloproliferative disorders) • Breakdown of screening their child will receive early on and a rationale for why it will be done	Degree of cognitive impairment can vary from mild (IQ 50-70), to moderate (IQ 35-50), or occasionally severe (IQ 20-35) and will reveal itself over time. Identify major medical problems their child may face and remove uncertainty by specifying what measures will be taken to treat or screen for these issues. Parents report resentment when information was seen as outdated, inaccurate, overly negative, or vague.
<i>* Defer conversations about conditions that may affect children with T21 later in life, unless they specifically ask for.</i> Ex. AML, ALL, celiac disease, seizures, obesity, Alzheimer, and depression <i>* If parents inquire or if you feel appropriate, highlight how children with T21 can live productively and well within today's society</i>	Children with T21 require lifelong screening so there is time for these issues to be addressed at a later date as parents report this can be overwhelming in the first conversation. This should be driven by parent interest but well known positive experiences such as involvement in the Special Olympics, work positions, and community roles may be discussed.
<i>* Re-evaluate parents' understanding to see if salient points have been communicated successfully</i> Ex. "We've covered a lot today. I want to make sure things were explained clearly. Please help me see if I left anything out"	By resetting expectations to frame questions as normal, you minimize feelings of embarrassment or inadequacy so that you can effectively provide a clear and robust explanation to their concerns.
<i>* Reiterate the specific steps in medical management will be taken in the next few days</i> <i>* Match the level of detail you provide about upcoming investigations and treatments to the readiness of the parents</i>	Remove uncertainty and clarify what will be done for their baby. Should encourage early involvement in child's care and belief that all necessary steps will be taken on their child's behalf.
<i>* Provide parents with information regarding local support groups and community initiatives</i> <i>* Supply parents with a handout containing information related to T21 and, if not available, provide a list of up-to-date electronic or print resources</i> <i>* Support parents by saying that they are not alone and you can come back for further clarifications or answering questions as needed</i>	Families should feel well-connected to healthcare supports as well as other families so they feel well taken care of and know they are not alone. Parents report feeling more optimistic when given current print materials so that they may research additional information at their own pace.
<i>* Conclude the conversation on a positive note</i> <i>* Plan a follow-up with them within the next 24 hours</i> <i>* Encourage parents to keep track of any questions that arise and reassure them that you will address them when you return</i>	Parents should be left with a sense of hope and an understanding that they are not alone. By coordinating follow-up, you provide time for them to process the information provided and give a direct demonstration of the ongoing support they will receive throughout their child's life.

Appendix E

Knowledge Based Principles - What Parents Want to Know ^{2, 8, 17}

- **What is Trisomy 21 and why did it happen to my baby?**
 - Genetic condition caused by presence of all or part of a third copy of chromosome 21 (also known as T21).
 - Approximately 95% of cases are due to spontaneous mutations (non-disjunction).
 - Generally, the recurrence risk is approximately 1% although this has some variability based on maternal age-related risk (i.e. higher risk of recurrence with increased maternal age).
 - About 3-4% of cases are due to chromosomal translocations with approximately 1/3 of these (i.e. 1% of all cases of Down syndrome) occurring due to hereditary inheritance patterns.
 - The remaining 1% of cases is due to mosaicism.
- **Why do you think my child has Down syndrome?**
 - Children with T21 are individuals that share many physical characteristics including: hypotonia; epicanthal folds; flat nasal bridges; upslanting palpebral fissures; speckling of the iris; abnormal auricles; hyperflexibility; excessive skinfolds in the posterior aspect of the neck; a single transverse palmar crease; and a wide gap between the first and second toes.
- **How common is Down syndrome?**
 - Estimates vary but the evidence shows that, overall, approximately 1 out of every 700 babies born is diagnosed with having Down syndrome.
 - Although the incidence increases with maternal age, the majority of children (i.e. approximately 80%) with T21 are born to women under 35 years old as this is in keeping with the baseline higher fertility rates.
- **Could I have prevented this?**
 - There is prenatal screening that can help to diagnose T21 before your child is born but, because most diagnoses are sporadic, there is nothing you could have done to prevent it from happening.
- **What is my baby at risk for right now?**
 - The most common conditions affecting neonates with T21 who are less than 1 month old include:
 - Hypotonia (correlates to feeding difficulties and aspiration risk)
 - Hearing problems (75%)
 - Congenital heart disease (40-50%)
 - Polycythemia (18-64%)
 - Feeding problems (correlates to the degree of hypotonia seen)
 - Cataracts (15%)
 - Gastrointestinal atresias (12%)
 - Constipation
 - Transient myeloproliferative disorder (10%)
 - Congenital hypothyroidism (1%)

- **What will be done for my baby in the first few weeks of life?**
 - Key points to mention include: a chromosomal analysis to confirm diagnosis; an echocardiogram to screen for congenital heart defects; a swallowing assessment for aspiration risk; an eye exam for cataracts; newborn hearing screen; history and exam for duodenal and anorectal atresia; baseline CBC for transient myeloproliferative disorders and polycythemia; typical newborn screening with particular emphasis on TSH levels to assess for congenital hypothyroidism; a discussion regarding risks of respiratory infections; and a discussion of recurrence risk.
- **What is the life expectancy of individuals with Down syndrome?**
 - Average life expectancy in Canada is currently into the mid-50's with 10% of adults living to be 70+ years.

Appendix F

Resources

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