Primary Care Provider's Guide to

WOMEN'S HEALTH AND DOWN SYNDROME

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LEAD AUTHOR, HANNAH GRAHAM, MD

Dr. Graham is a board-certified Family Medicine physician at the **Advocate Health Care Adult Down Syndrome Center** in Park Ridge, IL. The Center serves people aged 12 and older with Down syndrome. Besides her work at the Adult Down Syndrome Center, she is currently a faculty member with the Family Medicine Residency Program at Lutheran General Hospital and at Rosalind Franklin University Chicago Medical School. In her free time, she enjoys spending time with her family, which includes a young daughter with Down syndrome.

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Contributing Author (Cases 6 and 7), Terri Couwenhoven, MS, Certified Sex Educator (CSE)

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Down Syndrome Medical Interest Group (DSMIG-USA)
American Academy of Developmental Medicine & Dentistry (AADMD)

PROFESSIONAL REVIEWERS AND CONTRIBUTORS

Terri Couwenhoven, MS, Certified Sex Educator (CSE)
Lindsey Wilson, MD
Svea Welch, LPC-MHSP
George Capone, MD
Priya Chandan, MD, PhD, MPH
Brian Chicoine, MD
Laura Krohn
Katie Frank, PhD, OTR/L

SELF-ADVOCATE REVIEWERS AND CONTRIBUTORS

Charlotte Woodward, BA
Kayla McKeon, AA, LHD
Sabine Collins
Ashley Jones

E-LEARNING COURSE

Kramer Davis Health
Jamie Valis, PhD

Dear Reader,

As President and CEO of the National Down Syndrome Society (NDSS), it gives me great pleasure to introduce the NDSS *Primary Care Provider's Guide to Women's Health and Down Syndrome*. For more than four decades, NDSS has advocated for the rights, well-being, and inclusion of individuals with Down syndrome, and this guide is further evidence of that commitment.

Women with Down syndrome are living longer than ever. This expanded lifespan is a cause for celebration, while also introducing new challenges as these women often face unique barriers to accessing quality health care tailored to their specific needs. Estimates show that only 5% of adults with Down syndrome have access to a clinic that specializes in caring for adults with Down syndrome. Many rely solely on primary care providers for all their health care needs.

This guide aims to help providers better understand the specific health care needs of women with Down syndrome and provide insight into the Down syndrome community. By equipping health care professionals with knowledge and tools to provide tailored and compassionate care, we believe providers, like yourself, will improve health outcomes and enhance the quality of life for women with Down syndrome.

We are grateful to the WITH Foundation for their generous support of this guide, and we thank all the contributors, experts, and reviewers who have helped bring this guide to fruition.

Thank you for joining us on this important journey towards promoting women's health and fostering a more inclusive and understanding society. Together, we can make a meaningful difference in the lives of women with Down syndrome.

Sincerely,

Kandi Pickard
President and CEO

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DISCLAIMER: This guidebook was created for the sole purpose of educating physicians and other health care providers on the differences and similarities that exist in the health care of women with Down syndrome. This resource is not intended to be used by any person in lieu of the patient-specific recommendations of their health care providers. The information contained in this guidebook is wholly owned by the National Down Syndrome Society and may not be copied or otherwise used for any purpose other than the educational advancement of the reader. The National Down Syndrome Society is not, and shall not be, liable for any damages associated with the use or misuse of the information outlined in this guidebook.

INTRODUCTION

Down syndrome is the most common chromosomal condition diagnosed in the United States.[1] The mean life expectancy of a person with Down syndrome in 1950 was 26 years old; that mean has increased to 53 in the mid-2010s, although ethnic differences in life expectancy do persist, as a result of disparities in social determinants of health.[2] As the population of people living with Down syndrome grows, the understanding of the condition and its associated health differences continues to increase. In 2016, there were estimated to be 217,163 people with Down syndrome in the United States.[2]

Despite this increase in Americans with Down syndrome, it is estimated that only 5% of individuals aged 18 and older have access to a clinic specializing in caring for adults with Down syndrome.[3] Most people with Down syndrome receive medical care from their primary care physicians.

Additionally, one study estimated that women with Down syndrome received substantially less gynecological health care compared to women without Down syndrome, and women with Down syndrome received less gynecological care compared to other forms of health care.[4]

To help address the need for improved health care for women with Down syndrome, this guide provides primary care providers with current data and information about their unique health care needs. It is arranged into cases to demonstrate how clinicians may encounter these topics. The cases presented are not actual patients but are based on frequently met concepts and questions.

This guide is informed by women with Down syndrome. Their participation, comments, and questions provide an underlying current to every topic. Their self-advocate perspectives will be outlined throughout the guide. They send an unapologetically clear message: life with Down syndrome is great!

IMPORTANCE OF LEARNING ABOUT WOMEN'S HEALTH AND DOWN SYNDROME

Women with Down syndrome – just like all women and all people – have inherent value and worth and are deserving of good, comprehensive health care. However, health care providers may lack exposure to individuals with Down syndrome, leading to biases that impact their care. It's crucial for providers to educate themselves on women's health issues in this community to deliver informed and respectful care.

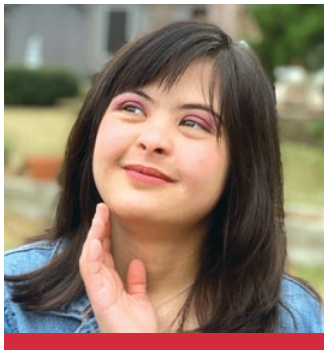
While women with Down syndrome may have some unique concerns relating to their health, they also share many of the same concerns as those of non-disabled women. Health care providers may need to adapt their communication with women with Down syndrome so that what is being discussed or done can be better understood by the patient. Sufficient time should be given to allow the patient to ask the questions they need to better understand their health issues and any procedures being conducted.

Additionally, some women, like me, may be accompanied by a family member or other companion/support personnel at appointments, but medical professionals should look at and speak directly to the patient. Doing so shows that they are presuming the competence of women with Down syndrome. Just like any other woman, I want health care professionals to be my allies and partners in my health care.

– Charlotte W.

Women with Down syndrome want to be treated with respect because we are people, just like anyone else – we fall in love, we get sad, we laugh, we cry. At times, we may require special attention to address our health needs, so it is important for doctors to know what is needed to keep us healthy and tell us how. Like how to eat right or how much sleep we need to get each night. Some of us may need a little extra help with certain tasks, but we strive to live our lives to the fullest and contribute to our communities in meaningful ways. With the right support and understanding, we can overcome challenges and achieve our dreams, just like anyone else, and doctors play a crucial role in helping us along this journey.

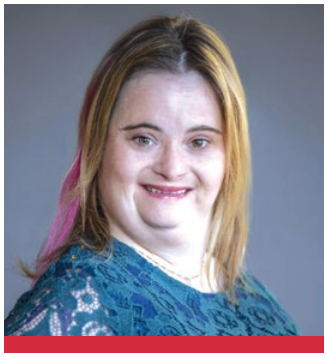
– Ashley J.



Doctors need to understand the specific tests required for women with Down syndrome and be aware of common health issues we may face. It is important for doctors to be prepared and knowledgeable about what's happening with women who have Down syndrome because we deserve the same level of care as women who don't have Down syndrome.

When providing care, doctors need to take the time to get to know each woman with Down syndrome, especially because we all have different health care needs. I always appreciate it when my doctor asks me questions about myself because it makes me feel like they care about me and my health. I think this is true for other women with Down syndrome too! When doctors take the time to listen to us, we feel more comfortable and supported in receiving care.

– Sabine C.



Like any woman, I have aspirations, concerns, and questions about my future and my health. I also have Down syndrome, but it is important to me that doctors recognize me as more than just my disability and approach me with the same respect, dignity, and attention to my health care needs as they would with any other patient. I believe that if doctors do more to understand the unique medical considerations and experiences of women with Down syndrome, they can be better at providing care that is tailored to each patient's specific needs and promote a more inclusive and supportive health care environment for all.

– Kayla M.

HOW WOMEN WITH DOWN SYNDROME LEARN

Women with Down syndrome display a wide range of intellectual disability and learning styles. Each woman should be thought of as an individual first with her own unique skills and needs. While a diagnosis of Down syndrome does not automatically confer certain needs, there are some general learning styles and cognitive functions that are quite common in people with Down syndrome.

In their book, *Mental Wellness in Adults with Down Syndrome: A Guide to Emotional Strengths and Challenges*, McGuire and Chicoine explain that people with Down syndrome are often concrete and literal thinkers.[5] Women with Down syndrome may do well with a concrete task at work or home that they are expected to accomplish in a literal and consistent way. However, a woman with Down syndrome may be so successful with accomplishing this task that she becomes inflexible performing outside of her concrete schedule or routine. Additionally, abstract thinking can be challenging for many people with Down syndrome. Questions around abstract concepts like appetite, time, and pain can be difficult for some women with Down syndrome to comprehend and answer.

A woman with Down syndrome may be able to answer a concrete question such as, "Do you wear a pad when you are on your period?" However, she might struggle to answer a question like "When did your last period start?" and might require the use of a calendar or app to make the dates of her period more concrete. She may also struggle to answer an abstract question like "Where is the pain?" and might be able to answer more concrete questions like "Do you have pain in your stomach?" more easily.

People with Down syndrome are sometimes described as having auditory processing deficits.[5] This means they have difficulty listening to an instruction and then completing the task. In contrast, many people with Down syndrome have strong visual memories. They remember things they see and often do well with visual instructions and supports. Using pictures to explain a concept or to map out a task that a person with Down syndrome needs to complete can be an effective tool. When appropriate, examples of these visual supports are included in this guide.

A general principle to keep in mind is that physical symptoms in women with Down syndrome can sometimes present as a change in behavior. Behaviors are a form of communication so determining what is causing the behavior requires an in-depth look at the situation.[5, 6] Sometimes, a behavior change may be related to a psychological concern. Sometimes, it is a change in routine or home life, and sometimes it is in response to physical discomfort or pain.[7] Often, clinicians and caregivers must work together with a patient to determine the root cause of these changes in behavior.

A Unique Learning Profile: Individuals with Down Syndrome

Hearing impairment

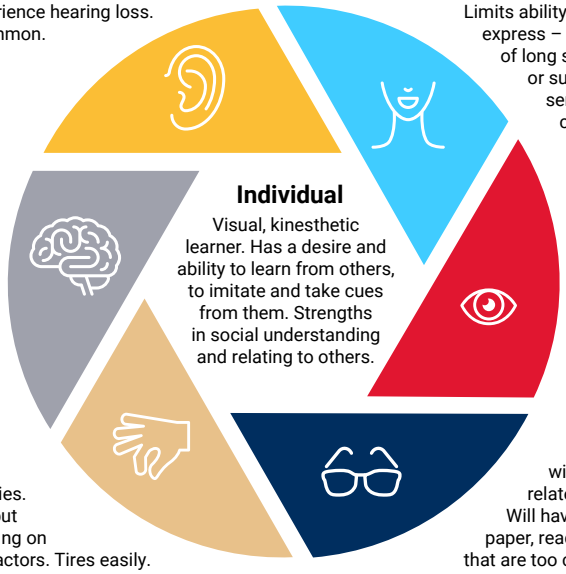
Some 50-70% of adults with Down syndrome experience hearing loss. Sensorineural and conductive hearing loss are common. Will have difficulty listening in noisy environments, processing spoken language, discriminating speech sounds, and learning phonics.

Verbal memory weakness

Difficulty learning from listening – maintaining attention, retaining instructions, memorizing sequences, and learning new vocabulary and information. Challenges in retaining and consolidating learning into long-term memory.

Delayed motor skills

Linked to low muscle tone, loose ligaments, and developing motor plans. Affects all physical activities. Delayed self-help skills and handwriting progress but will improve with practice. May have difficulty staying on task and multi-tasking. Easily distracted by other factors. Tires easily.



Speech & language delay

Limits ability to communicate. Understands more than can express – knowledge may be underestimated. Affects processing of long sentences, learning from listening, understanding new or subject-specific vocabulary, word finding, forming sentences, understanding instructions, reading comprehension skills, and thinking and reasoning.

Visual learning strengths

Ability to learn and use signs and gestures, to learn to read and use written words. Strengths in learning through imitation, from modeling and demonstration. Learns well from visual resources (pictures, photos, diagrams, symbols, concrete materials, digital technologies, and apps).

Visual impairment

Common in adults with Down syndrome. Challenges with depth perception. Can experience early onset of age-related vision issues. Bifocals are routinely recommended. Will have difficulties with writing, using a pencil on blue-lined paper, reading <18 point font, coping with text/diagrams/pictures that are too cluttered, detailed, or have little contrast.

This graphic, adapted from *Down Syndrome: Good Practice Guidelines for Inclusive Education,* outlines strengths and challenges associated with the main aspects of the specific learning profile for people with Down syndrome, as well as the unique range of sensory, physical, and cognitive needs of this group of learners.

CASE 1

Emma is a 13-year-old with Down syndrome who presents to her primary care provider's office with questions about her menstrual cycles. Emma first started menstruating at age 12. Her periods are regular and last four to five days. Emma is usually independent with toileting, but she is struggling to manage her period hygiene, requiring assistance from her mother to change pads at home and having accidents where blood leaks onto her clothes at school. As a result, she does not want to go to school on days when she has her period. Emma and her mother are looking for suggestions to help Emma with her period.

MENARCHE

In the United States, age of first period (menarche) is around age 12.[8] There is very little published literature about the age of menarche in adolescents with Down syndrome, and some of the literature is decades old. One more recent review found that adolescents with Down syndrome typically underwent menarche around the same age or slightly earlier than adolescents without Down syndrome.[9] A trend toward slightly younger age of menarche in adolescents with Down syndrome may be related to higher obesity rates, but this has not been proven and some studies have not found this association.[9]

PERIOD HYGIENE

While some women with Down syndrome continue to require assistance with managing period hygiene, many adolescent and adult women with Down syndrome can be fully or partially independent in managing period hygiene with concrete teaching, advance preparation, adequate support, and practice. Pharmacologic interventions to make periods stop needn't be the first line of treatment for period hygiene as these hormonal medications can have side effects. Many women with Down syndrome can learn to manage their periods without hormonal interventions.

There are several strategies that can help develop independence in period hygiene for women with Down syndrome.

Because people with Down syndrome tend to be strong visual learners, creating a visual story around period hygiene can help women with Down syndrome learn how to manage periods. The visual supports can be individualized to the woman to explain what menstruation is and how to manage menstrual pads. Using a real pad for practice when a woman is not on her period can be helpful, and using a red marker or food coloring to color a practice pad can also help reinforce some of the visual cues about changing pads. Visual supports for period hygiene are available.[10] Examples and resources can be found at the end of this guide.

For women learning to manage period hygiene, creating a schedule with reminders for when to change a pad can be helpful. Those reminders could come from a caregiver or by setting a timer to go off on a phone every few hours. Some women with Down syndrome learn to change their pad every few hours, and some women learn to change their pad every time they go to the restroom while menstruating. Strategies can be individualized to help encourage maximum independence.[11]

Keeping a bag of period supplies (a period kit) that a woman may take with her when she uses the restroom while menstruating can be helpful. The kit can contain pads, wipes, and an extra set of underwear and pants in case of leaks.

Some women with Down syndrome find the fine motor movements of using a pad to be challenging. Using different pads can help with this. Using extra-long pads and pads without wings can be easier. There are also reusable pads available in different brands.

SELF-ADVOCATE PERSPECTIVE: MENSTRUATION

Menstruation is the same for all girls and women. All the women in my family have started their menstrual cycle at about the same age. However, unlike my sisters, my teacher and mom got together to discuss how to support me. The teacher and her aide made a book of pictures showing what to do each month. At the start of the month, we would do a calendar count and make sure that I had extra clothes at school. With time, I've grown more independent and confident, and can now manage the process by myself.

All girls need to learn how to take care of their menstrual needs along with what is best to wear in case of an accident, so they won't get embarrassed.

Doctors can help women with Down syndrome better understand what menstruation is by explaining it and by creating or finding visual supports, like the one my teachers made me, to help patients remember the steps and know what to do.

–Ashley J.

In the last decade, period-absorbent underwear has become available. This type of underwear absorbs menstrual blood like a pad, but it is washable and reusable. Period-absorbent underwear or "period panties" can be used on its own or in addition to a pad when menstrual bleeding is heavier. Because period-absorbent underwear offers protection from leaks, it can be a helpful addition for women managing period hygiene. There are many brands and styles available for purchase. There are also period-absorbent swimsuits to accommodate menstruation while swimming.

Women with Down syndrome may be interested in learning to use a tampon to manage menstruation, and some women have successfully learned to use one, often being taught by their mothers or sisters. However, it is important to keep in mind that learning to use a tampon can require a lot of trust between the woman with Down syndrome and her teacher. Additionally, because women with Down syndrome are at higher risk of sexual abuse, learning to use a tampon should be an individual decision made with supportive caregivers/teachers and with appropriate sexual health safety measures in place. Women who desire to use a tampon should be taught to insert the tampons themselves, rather than having a caregiver insert the tampon. With the availability of period-absorbent swimsuits, many women with Down syndrome may be able to enjoy recreational swimming without needing to master tampon usage.

Emma and her mother are interested in trying some of the strategies to help Emma become more independent in managing her period. They state that Emma's sister just received an HPV vaccine, and they wonder if Emma will need this vaccine.

HPV VACCINATION

Human Papillomavirus, or HPV, is a major cause of cervical cancer, vulvar cancer, vaginal cancer, oropharyngeal cancer, and genital warts.[12] A 9-valent vaccine against HPV has been found to be safe and effective at preventing HPV infection and is recommended for women within designated age groups.[12,13] There are no specialized recommendations regarding HPV vaccination for women with Down syndrome. However, one study found that women with Down syndrome were less likely to receive HPV vaccination.[14]

Because HPV is a sexually transmitted disease, women with Down syndrome and their families often consider sexual activity when deciding on the vaccine. Some women and their families assume that they or their loved one with Down syndrome will never be sexually active and, therefore, decline the HPV vaccine. While some women with Down syndrome do choose to become sexually active in romantic relationships, all women with intellectual disabilities (including Down syndrome) are at increased risk of sexual abuse.[15,16] Therefore, because the vaccine is safe and effective at preventing HPV infection, it seems reasonable that the HPV vaccination should be recommended for women with Down syndrome just as it is for women without Down syndrome. More research in this area is needed.

Emma and her mother are surprised to hear that HPV vaccination is also recommended for Emma. They are interested in pursuing this vaccine series.

CASE 2

Asha is an 18-year-old with Down syndrome who presents to her primary care provider with heavy and painful periods. Asha has monthly periods that last eight days and seem very heavy. She will need to change her pad every two hours for the first three days of her period. Day four of her period is usually lighter until it ends on day eight. Besides the heavy bleeding, Asha experiences tearfulness and seems to have cramps and pain the day before and the first three days of her period. She will often vomit and has missed school because of the pain. She becomes very irritable at the start of her period and seems uncomfortable. She is not sexually active. Asha and her mother are wondering if hormonal or pain medications would help her heavy and painful periods. They also wonder if she can take medication to stop her periods.

MENORRHAGIA

Menorrhagia is defined as excessive menstrual bleeding lasting more than seven days causing disruption to quality of life.[17, 18] The incidence of menorrhagia in women with Down syndrome has not been reported. The diagnostic approach to a woman with Down syndrome experiencing menorrhagia should be like that for women without Down syndrome. Because thyroid disorders are more common in women with Down syndrome, it is reasonable to check thyroid function tests in the setting of menorrhagia. It is worth noting there are no specific studies evaluating thyroid disorders causing menorrhagia in women with Down syndrome. Additionally, because of increased risks of leukemias, leukopenia, and other blood disorders in women with Down syndrome, checking a complete blood count can prove useful.

Just as in women without Down syndrome, blood labs, endometrial biopsy, transvaginal ultrasound, saline infusion sonohysterogram, or hysteroscopy may be indicated.[17] Many women with Down syndrome find it very difficult to

tolerate pelvic exams, and fewer women with Down syndrome can tolerate endometrial biopsies or transvaginal ultrasounds. In some cases, an abdominal ultrasound may supply enough information and potentially avoid a transvaginal ultrasound. When a woman with Down syndrome is not able to tolerate a necessary pelvic exam, biopsy, or imaging study, the care team can consider performing these exams or studies under general anesthesia. Weighing the need for the tests with any risks surrounding anesthesia is an important discussion between the woman with Down syndrome, her caregivers, and the medical team. Sometimes bundling this with other testing the individual finds difficult (such as ear cleanings, dental procedures, and blood draws) can optimize the use of anesthesia.

DYSMENORRHEA

Dysmenorrhea, or painful menstruation, can be associated with cramping pain in the lower back, abdomen, and thighs. It can be accompanied by nausea, vomiting, food cravings, and breast tenderness. For many women with Down syndrome, describing these specific symptoms can be difficult. They may instead present with irritability, which is very common, or other behavioral changes around the time of menstruation.

Dysmenorrhea is common in the general population, and while there appear to be no studies on the incidence of dysmenorrhea in women with Down syndrome, it seems to be common in women with Down syndrome as well.[19] The treatment and workup for primary and secondary dysmenorrhea are the same for women with Down syndrome as for women without Down syndrome. For primary dysmenorrhea, nonsteroidal anti-inflammatory drugs (NSAIDs) are a first line for treatment and work by decreasing prostaglandin production.[20] They have been shown to be more effective than acetaminophen and placebo at reducing symptoms of dysmenorrhea.[20] Because irritability is a common presentation of dysmenorrhea in women with Down syndrome, some women start taking NSAIDs around the time when they expect their period to start in order to prevent the symptoms from starting or worsening. While women with Down syndrome can experience GI side effects from NSAIDs, NSAIDs can be useful treatments with intermittent use and attentive monitoring. In addition, hormonal therapy is also considered first line for treatment of dysmenorrhea. Due to concern for side effects, it might be reasonable to consider an initial trial of NSAIDs prior to starting hormonal therapy.

Besides NSAIDs and prescription hormonal therapy, there is evidence supporting the nonpharmacologic treatment of dysmenorrhea. Exercise and heat (hot water bottles/heating pads) have been shown to possibly reduce symptoms (although this likely needs to be confirmed with more high-quality studies).[21, 22] Encouraging water intake, eating healthy foods, and wearing loose-fitting clothing (bras/shirts/pants) can help a woman feel more comfortable.

Asha and her mother decide to try ibuprofen to improve her cramps, pain, vomiting, and irritability. Asha's labs reveal a TSH within the normal range and a CBC with normal hemoglobin and platelet levels. After three months, Asha and her mother return to her primary care provider. She continues to exhibit irritability around her periods and has missed school because of her pain and nausea. Since her last visit, Asha and her mother have considered hormonal treatments for her symptoms and are interested in learning about these and other forms of birth control to prevent unintended pregnancy.

BIRTH CONTROL

Oral hormonal contraceptive pills (OCPs) are commonly prescribed for women without Down syndrome. Combined estrogen-progesterone oral contraceptives (COCs) are generally well tolerated. When deciding on OCP use for women with Down syndrome, the usual considerations around side effects and contraindications should be considered. There are also some unique considerations for women with Down syndrome when considering OCP use.

In women without Down syndrome, the most common side effects are breakthrough bleeding, breast tenderness, nausea, abdominal cramping, headaches, and increased vaginal discharge.[23] COCs can increase the risk of blood clots, stroke, elevated blood pressure, and myocardial infarction.[23]

People with Down syndrome appear to have lower rates of hypertension, although there are no studies evaluating blood pressure in women with Down syndrome on COCs.[24] There is some concern that women with Down syndrome may be at increased risk of blood clots. One retrospective analysis looked at children with Down syndrome and found that Down syndrome may be an independent risk factor for thromboembolism.[25] Additionally, people with Down syndrome are at higher risk of other autoimmune conditions, which are associated with increased risk of blood clots.[26] The risk of blood clots for people with Down syndrome needs further research. Because of this possible increased risk of blood clots in women with Down syndrome and the other risks associated with COCs, it is important to discuss the risks and benefits of treatment with COCs.

COCs are associated with lower rates of some cancers, including endometrial and ovarian cancers.[27] However, there is a possible increased risk of cervical and breast cancer in women who have used COCs.[27] It appears women with Down syndrome have overall lower rates of solid tumor cancers including breast and cervical cancer.[28]

One contraindication for COCs is migraine with aura.[23] Because it can be difficult for some women with Down syndrome to describe or express whether they have an aura with their migraine, it may be best to avoid COCs in women with Down syndrome who experience migraines and cannot definitively express a lack of aura symptoms.

Congenital heart disease is another consideration in initiating COCs. The *GLOBAL Medical Care Guidelines for Adults with Down Syndrome* recommend ongoing monitoring by a cardiologist for people with Down syndrome who have congenital heart disease.[29] There is a concern for a higher risk of embolic strokes in this population, and valvular heart disease is considered a contraindication for COC use in women without Down syndrome.[23, 29, 30] For women with Down syndrome and a history of congenital heart disease, consultation with a cardiologist before initiating a COC may be necessary.[31]

Progesterone-only pills (POP) are another hormonal contraceptive option. They are associated with irregular (breakthrough) bleeding and elevated blood pressure. They can also impair glucose metabolism for the first six months of use.[23] Most studies do not demonstrate an increased risk of blood clots associated with most formulations of POPs.[32] Because they are not associated with blood clots, POPs may be a more attractive option for women with Down syndrome. It is worth noting that POPs are a category 1 (no restrictions) method for those with valvular heart disease.[18] However, they must be taken at consistent times and the breakthrough bleeding can be difficult for women with Down syndrome to manage. A risk/benefit discussion with the patient and caregivers is necessary.

Injectable progesterone is another hormonal contraceptive option. Because it is an injection every few months, it is an attractive option for many women who do not want to take a daily pill. Long-acting injectable progesterone has similar side effects as oral progesterone. However, some studies showed an association with an increased risk of blood clots.[32] Injectable progesterone (e.g., depot medroxyprogesterone acetate) is associated with a loss of bone mineral density, although there is evidence to suggest improvement in bone mineral density occurs upon stopping the injectable.[33] Weight gain is also more common when using an injectable progesterone. Because osteoporosis may be more common in people with Down syndrome, this should be taken into consideration before starting an injectable progesterone.[28]

Seizure disorders are more common in people with Down syndrome.[28] Generally, women with epilepsy may have special considerations for management of their contraceptive needs. Because there can be drug-to-drug interactions between anti-epileptic medication and hormonal contraceptives, management of these medications in collaboration with a neurologist is helpful.[34]

A levonorgestrel-releasing intrauterine device (IUD) is another common contraceptive prescribed in women without Down syndrome. They are not associated with increased risks of blood clots, but they can be associated with irregular bleeding/spotting.[32] Many women stop having menstrual bleeding while the levonorgestrel-releasing IUD is in place. Placing an IUD is usually an in-office procedure involving a pelvic exam. Women with Down syndrome may not tolerate this exam. Although no research was found to corroborate, clinical experience has found that some women with Down syndrome who have had an IUD placed under anesthesia have reported satisfaction with their IUD. Rarely, IUDs may cause pelvic, cervical, or vaginal discomfort. In these scenarios, some women with Down syndrome may not be able to express this discomfort. Considering a woman's ability to express any side effects from an IUD may be an important consideration before selecting this method.

Implantable devices are small, soft rods that are inserted under the skin of a woman's arm. They are an in-office medical procedure and provide a continuous supply of progesterone. The in-office procedure might be difficult for some women with Down syndrome to tolerate. Implantable devices can cause breakthrough bleeding. A systemic review found that they did not have a statistically significant increase in risk of blood clots.[32]

Non-progesterone IUDs are hormone-free and work to prevent pregnancy. They can be associated with heavier and more painful periods. Like levonorgestrel-releasing IUDs, they are placed in an in-office procedure with a pelvic exam, which may be difficult for some women with Down syndrome to tolerate. While non-progesterone IUDs are effective forms of birth control, their side effects of heavier and more painful periods along with difficulty tolerating placement make them unlikely to be the preferred method for most women with Down syndrome.

Non-medication forms of birth control consist of natural family planning and barrier contraception methods. These methods require close self-monitoring and/or manual dexterity, which may be difficult for some women with Down syndrome to manage. They also have high failure rates in couples without Down syndrome, making them an unlikely solution for women with Down syndrome.[35]

PERIOD SUPPRESSION

Period suppression refers to the practice of stopping or lessening menstrual periods with the use of hormonal treatments. Hormonal contraceptive options can be used in this capacity. This practice is considered medically safe.[36] It is not recommended to start this practice before menarche.[36] While learning about menstruation can be challenging for women with Down syndrome, with education and practice, many women can become independent in period management and more tolerant of menstruation in their lives. Respect for the woman with Down syndrome's ability and willingness to learn about menstruation and support for maximizing her autonomy are important considerations before initiating period suppression. It may be a helpful option for some women with Down syndrome but should be initiated only after a discussion with patients and caregivers around realistic expectations for period management and the risks and benefits of medications.

SELF-ADVOCATE PERSPECTIVE: BIRTH CONTROL

I haven't yet explored birth control options because I'm not sexually active. However, I am interested in learning more because I've heard it can stop PMS cramps, which is something I struggle with. From my research and conversations with my mom, I've learned that there are many different types of birth control, which can include getting your tubes tied, intrauterine devices (IUDs), implants, shots, and the pill. Out of these, I think the pill might be a good choice for me because it can help with my cramps and bleeding, but I still think it's important for doctors to explain everything to me so I can choose what's best for my health. It's important for women with Down syndrome to have the same information and choices as everyone else.

–Sabine C.

Asha and her mother discuss the different hormonal contraceptive options to treat her painful periods. They understand that Asha may be at increased risk of blood clots. However, because her symptoms are so disruptive to her life, they feel that the benefits of starting a medication outweigh the risks. They do not think that Asha would tolerate a pelvic exam and would like to avoid having her undergo an invasive procedure for now. They decide to start a COC pill. The provider counsels on the side effects of the medication, including increased blood pressure. Asha's blood pressure tends to run on the low side, which is common in people with Down syndrome.[37] They plan to start the medication and follow up in three months for continued monitoring.

CASE 3

Yasmin is a 19-year-old with Down syndrome who presents to her primary care provider's office with irregular periods. Her periods have always been irregular and have become more sporadic over the last year. Recently, Yasmin has skipped her period for several months at a time. She will often go two to three months without a period. She has also noted an increase in facial hair growth. She is not known to be sexually active.

OLIGOMENORRHEA AND AMENORRHEA

Oligomenorrhea refers to menstrual cycles which are between 35 days and six months in length.[38] Primary amenorrhea refers to not reaching menarche by 15 years of age or three years after thelarche.[38] Secondary amenorrhea refers to the cessation of periods for three months after having regular periods or six months after having irregular periods.[38] There are no studies evaluating the frequency of oligomenorrhea and amenorrhea in women with Down syndrome. In women without Down syndrome, both conditions require further testing, and women with Down syndrome should probably undergo testing similar to women without Down syndrome. Pregnancy should be ruled out in cases of oligomenorrhea and amenorrhea. As pregnancy is uncommon but not impossible in women with Down syndrome, it does need to be considered. Additionally, oligomenorrhea and amenorrhea may be the symptom of many endocrine disorders. Endocrine disorders are more common in women with Down syndrome, so further testing is warranted.[39]

POLYCYSTIC OVARIAN SYNDROME (PCOS)

Polycystic ovarian syndrome (PCOS) is common and affects 7% of reproductive-aged females in the United States.[38, 40] One study looking at PCOS in women with Down syndrome found that, although endocrine disorders are more common in people with Down syndrome, PCOS was less common in women with Down syndrome compared to the population as a whole.[39] Even though PCOS may be less common in women with Down syndrome, there are likely still many women with Down syndrome with a diagnosis of PCOS because PCOS is so common.

Diagnostic criteria for PCOS vary depending on the organization but many organizations recommend the 2003 Rotterdam Criteria.[40] The Rotterdam Criteria require a woman have two of the following three symptoms: androgen excess, ovulatory

dysfunction, or polycystic ovaries. Symptoms of androgen excess like hirsutism, acne, and androgenic hair loss can be easily observed in women with Down syndrome. Attention to menstrual frequency can also indicate signs of ovulatory dysfunction. However, polycystic ovaries can only be diagnosed via imaging (preferably transvaginal ultrasound). A transvaginal ultrasound may be difficult for many women with Down syndrome to tolerate. In women who are not able to tolerate this exam, sometimes a transabdominal ultrasound can supply enough information about the status of the ovaries.

Prior to a diagnosis of PCOS, it is recommended to rule out other common disorders, including pregnancy, thyroid disease, hyperprolactinemia, and nonclassical congenital adrenal hyperplasia.[40] There are more rare conditions that may be appropriate to rule out depending on the presentation.[40] Generally, the typical evaluation for PCOS is appropriate for women with Down syndrome, particularly since women with Down syndrome have an increased prevalence of some endocrine disorders.[39] There are also common comorbidities in women who have PCOS which may be more common in women with Down syndrome, including mood disorders and sleep apnea.[38, 28]

Treatment of PCOS is classically affected by whether pregnancy is desired. Since pregnancy is not desired for most women with Down syndrome, treatment with first line medications like hormonal contraceptives or metformin may be initiated after discussion of risks and benefits with the patient and caregivers.[38] Second-line treatments for PCOS can be tailored to specific patient scenarios and may include spironolactone or topical treatments for hirsutism and acne.

Yasmin agrees to undergo blood tests because of her irregular periods and hair growth. Although she is not known to be sexually active, they agree to a pregnancy test because women with intellectual disabilities are at risk for abuse. She and her caregivers decline a transvaginal ultrasound because they do not feel she would tolerate the exam.

Her pregnancy test is negative and her blood tests are normal. Because her facial hair growth is bothersome to her, she is interested in starting a medication to treat this. However, she and her caregivers are concerned about her risk for blood clots. After a discussion of options, risks, and benefits, they would like for Yasmin to start on spironolactone to treat her hirsutism. As her blood pressure often runs on the low side, they will monitor her blood pressure closely on the spironolactone. They are also interested in starting metformin to treat her irregular periods.

CASE 4

Keisha is a 22-year-old with Down syndrome who presents to her primary care provider with irritability and worsening behaviors around her period. Keisha just moved to a new group home six months ago. She is nonverbal and is observed to have outbursts where she will grab at her housemates and refuse to participate in preferred activities. She prefers to stay alone in her room, and it is difficult to convince her to get out of bed. These behaviors seem to be worse in the week before her period and resolve after her period is over.

PMS AND PMDD

Premenstrual syndrome (PMS) and premenstrual dysphoric disorder (PMDD) are common. Up to 12% of women experience these disorders.[41] There are no studies comparing the prevalence of these conditions in women with Down syndrome. They appear to be as common in women with Down syndrome as in women without Down syndrome.[19]

PMS is a constellation of symptoms experienced during the five days before menstruation for at least three consecutive cycles.[41] The diagnosis requires the presence of at least one mood symptom and one somatic symptom that begin during the luteal phase. These symptoms should resolve within four days of onset of menstruation and not recur until at least day 13 of the next cycle. Symptoms should be present in the absence of hormone, alcohol, and drug use and should impact social, academic, or work performance to be defined as PMS.[41]

PMS symptoms can include angry outbursts, anxiety, confusion, depression, irritability, social withdrawal, abdominal bloating, breast tenderness or swelling, headache, joint or muscle pain, swelling of extremities, or weight gain.

PMDD is a more severe form of PMS. To meet diagnostic criteria for PMDD, a woman should have at least five mood and physical symptoms. She must have one or more of the following: lability; irritability, anger, or increased interpersonal conflicts; depressed mood or feelings of hopelessness; anxiety, or tension. She must also have one or more of: decreased interest in usual activities, difficulty in concentration, lack of energy, change in appetite, insomnia or hypersomnia, feeling overwhelmed, and/or physical symptoms like breast tenderness, joint or muscle pain, or bloating. In addition to having one symptom from each category,

she should have a total of five symptoms. These symptoms must be present in the week before her period starts and stop within the first few days of the start of her period. They should remain absent for a week after completing menstruation. These symptoms should be present with most menstrual cycles for a year to be diagnosed as PMDD.[41, 42]

It is sometimes difficult for women with Down syndrome to express their discomfort while experiencing these symptoms. In such cases, the diagnosis is sometimes made when a caregiver notices irritability and changes in behavior related to the timing of the menstrual cycle of the woman with Down syndrome. The differentiation between PMS and PMDD in these cases might depend on the severity of a woman's symptoms or how much they are impacting her functioning.

Treatments for PMS and PMDD usually address the body's hormonal systems or neurotransmitters.[41] Studies suggest that OCPs are beneficial in treating PMS and PMDD. Women with Down syndrome may need special considerations before starting OCPs, including understanding the risks for side effects or other contraindicated conditions (as discussed above).

Selective serotonin reuptake inhibitors (SSRIs) have FDA approval for the treatment of PMS and PMDD in the United States. They can be taken during the luteal phase or continuously to treat symptoms.[43] They have been shown to be effective at treating psychological symptoms as well as irritability and physical symptoms.[43] It is generally easier for women with Down syndrome to take these medications on a regular basis rather than trying to time them with menstrual cycles. Because some women with Down syndrome may have trouble expressing side effects related to the medications, it is sometimes helpful to start at lower doses and to slowly titrate doses up as needed.[5]

SELF-ADVOCATE PERSPECTIVE: PMS

Since starting my period, I have experienced Premenstrual Syndrome (PMS). My emotions change a lot right before my period. Sometimes I feel irritable, other times I feel energetic, and sometimes I even feel depressed. When PMS hits, a lot of thoughts run through my head, and it causes me stress. There are even times when I become lethargic and just want to stay in my bed all day. Like some other women, I get very painful cramps that make me feel nauseous. Thank goodness these cramps only last the first day, but they are still terrible!

It's important for doctors to help me, and other women with Down syndrome, during this time. Doctors can give us advice on how to feel better, whether it's with medicine or other things, like a heating pad. This information can be so helpful and make PMS a bit more manageable!

–Sabine C.

After a discussion with Keisha, her care team, and her guardian, the decision is made to start sertraline 25 mg to treat her symptoms. After two months, her staff reports that she is still irritable around her periods and does not want to participate in all her normal activities, but she is getting out of her bed and is no longer grabbing at housemates. The decision is made to increase her sertraline to 50 mg.

After taking the sertraline 50 mg for two months, Keisha shows further improvement in her symptoms around her periods. Her irritability is markedly improved, and she participates in all her usual activities.

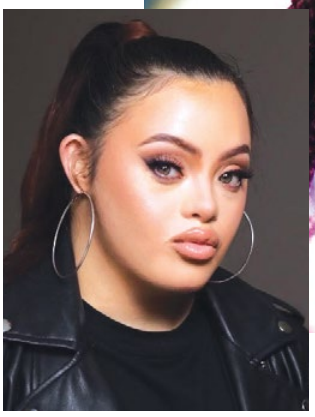
CASE 5

Jessica is a 25-year-old with Down syndrome who presents with her sister to her primary care provider for a well-woman exam. She has never had a pelvic exam and is not known to be sexually active. Her periods are regular, last four to five days, and are not too heavy. She has received the HPV vaccination series. She and her sister wonder if she should have a pap smear done.

CERVICAL CANCER

While people with Down syndrome are at increased risk of lymphoma and leukemia, they are at decreased risk of most solid tumor cancers including breast and cervical cancer.[28, 44, 45] Cervical cancer is less common in all women with intellectual disabilities.[46, 47] Risk factors for cervical cancer, such as smoking and multiple sexual partners, are presumed to be lower in women with Down syndrome than in the general population.[45] In a study of solid tumors in people with Down syndrome, there were no cases of cervical cancer reported, and in a study of cancer mortality in people with Down syndrome, no mortality due to cervical cancer was reported. [44, 48]

Despite this lower risk of cervical cancer, it is important to underscore that women with intellectual disabilities are at risk of sexual abuse.[49] Therefore, their exposure to HPV, a major risk factor for cervical cancer, may be unknown. Although rare, cervical cancers have been discovered at a very advanced stage in women with Down syndrome.[50]



There are no formal cervical cancer screening guidelines specific to women with Down syndrome. The American College of Obstetricians and Gynecologists (ACOG) continues to recommend cervical cancer screening for women aged 21-65 regardless of sexual activity.[51] This screening would aim to have a high level of sensitivity and not miss a case of cervical cancer in a woman who was not known to be sexually active. For women with an intellectual disability, some propose cervical cancer screening being tailored to her risk for cervical cancer.[52] Similarly, for women with Down syndrome, some propose beginning cervical cancer screening at age 21 and developing an individualized approach for the timing of repeat pap smears.[15] Pelvic exams and HPV testing can be very difficult for some women with Down syndrome to tolerate. As such, there are many approaches and variations in practice due to the lack of consensus around cervical cancer screening in women with Down syndrome.[45]

Jessica's primary care provider explains that the major risk factor for cervical cancer is HPV exposure. Jessica is not known to be sexually active, although she is at higher risk of sexual abuse in which case her HPV exposure may be unknown. The provider also explains that women with Down syndrome have historically very low rates of cervical cancer. The provider reviews that there are no formal guidelines specific to cervical cancer screening for women with Down syndrome and that, in the United States, ACOG recommends cervical cancer screening without consideration of sexual history. Because Jessica and her sister feel that her risk of cervical cancer is very low, they choose not to have her undergo a pap smear for cervical cancer screening. They think a pelvic exam would be very difficult for Jessica, but the main reason behind their decision to forego cervical cancer screening is because of her very low risk of cervical cancer.

PELVIC EXAMS

A pelvic exam can be difficult for some women with Down syndrome to tolerate. Preparing the woman for the exam with visual supports can be helpful. An example of a pelvic exam social story is available to review with the woman before the exam (see resources).[53] Additionally, having a trusted female relative or caregiver with the patient in the room can be helpful. Even if the provider is female, it is recommended to have a female nursing staff in the room as well.

SELF-ADVOCATE PERSPECTIVE: HPV VACCINE AND CERVICAL CANCER SCREENING

As I have learned more about my reproductive health, I think it is important to protect myself from any potential infections, so I think it makes sense for women with Down syndrome to take preventative measures to protect their health such as receiving HPV vaccinations and routine cervical cancer screenings. Some women with Down syndrome might not have learned about these things, so health care professionals should explain the benefits of HPV vaccination to women in a way that is easily understood, and they should also answer any questions they might have.

I feel respected during medical appointments when my questions and concerns are acknowledged by my gynecologist, and they provide thorough answers and explanations. Some women with Down syndrome may feel more comfortable with gynecological screenings if they have a female doctor or gynecologist doing these important procedures.

–Charlotte W.

Modifications to the exam may lead to higher success rates. Avoiding use of stirrups and having the woman use alternate positions, such as pulling her thighs to her chest or putting her feet together and spreading her knees apart, can help make the exam more tolerable. Some women do not tolerate the speculum exam, in which case using a one-finger exam to touch the cervix and then using that finger to guide the brush to collect cells from the cervix can be done. In other cases, a blind pap smear, performed by placing the brush in the vagina towards the cervix without visualizing the cervix, may need to be performed. The bimanual exam may need to be performed with one finger, especially in women with an intact hymen or small introitus. *A Health Handbook for Women with Disabilities* includes depictions of various pelvic exam positions.[54]

BACTERIAL VAGINITIS AND VULVO-VAGINAL CANDIDIASIS INFECTIONS

One study showed a lower rate of bacterial vaginosis in women with Down syndrome, but no studies were found that investigated prevalence rates of vulvovaginal candidiasis infections in women with Down syndrome. Women with Down syndrome may be less likely to tolerate topical or suppository treatment formulations. In women who will not tolerate those applications, oral agents may be necessary.[55]

STI TESTING

One study was found that reported on sexually transmitted diseases, and most were statistically less common in people with Down syndrome.[55] It is important to keep in mind that women with Down syndrome have lower rates of sexual activity but higher rates of sexual abuse. As such, screening may need to be tailored to a woman's symptoms and individualized after risk and benefit discussions with the patient and her caregivers. In women with Down syndrome who do choose to become sexually active, it is important to provide education for STI prevention.

Jessica undergoes a pelvic exam with a blind pap smear. At that time, swabs for STI and vulvovaginal testing are obtained. When all the results are reported a few days later, she has a normal pap and STI testing. She appears to have a yeast infection. Her sister reports she does not want Jessica to have to use topical treatments as Jessica is still upset after her recent pelvic exam. Jessica's sister opts for her to undergo oral treatment. A few weeks later Jessica's sister calls to report Jessica is feeling better, both from her yeast infection and from undergoing the pelvic exam.

SELF-ADVOCATE PERSPECTIVE: PAP SMEARS

It is important that women with Down syndrome receive comprehensive health care, including reproductive health care. Women with Down syndrome may be anxious about or fearful of medical appointments and procedures that involve exposing and examining private parts of their bodies. Doctors need to acknowledge this and be mindful of the disability community's concerns about reproductive health care procedures due to past harmful practices, such as the forced sterilization of women with disabilities.

Health care professionals should try to make the experience as comfortable as possible by explaining what they are going to do before they do it, by showing the patient the tools they are going to use, and by using appropriately sized tools. What helped me was accompanying my mom during an appointment so that I could understand and become familiar with the process. Learning about pap tests, why they are important, and what to expect ahead of time may make women with Down syndrome feel more comfortable when they are having this procedure done.

–Charlotte W.

CASE 6

Kara is a 27-year-old with Down syndrome who presents with her sister, her legal guardian, to her primary care provider for a well-woman exam. During the appointment, Kara reports she now has a boyfriend whom she has been dating for the past six months. Her sister expresses concern about Kara understanding her rights in a dating relationship and worries about the risk of exploitation.

SEXUAL EXPLOITATION

Sexual abuse is defined as unwanted or coerced sexual activity (genital touching, sexual activity, or intentional exposure to genitals or graphic media). Although there is no research specifically on individuals with Down syndrome, there is considerable documentation of the increased incidence of sexual abuse in people with intellectual disabilities.[49] It is estimated that one in three adults with intellectual disabilities have experienced sexual abuse.[49] For women with Down syndrome, the physical signs and symptoms of abuse are similar to the rest of the population. However, the ability or willingness to report the abuse may be limited and the incidence of underreporting is thought to be greater.[49, 56] Observation of behavioral or psychological symptoms and consideration of abuse as a possible cause is important. These changes may include self-injurious behavior (SIB), inappropriate sexualized talk, anxiety about a particular place or person, and depression. PTSD may also be exhibited. If the family reports sudden changes in behaviors linked to a particular place, event, or person, the possibility of sexual exploitation should be considered.

Kara and her sister are saddened to hear that women with Down syndrome are at higher risk for sexual abuse. During this discussion, Kara reveals her boyfriend has raped her.

DISCLOSURE

Women with intellectual disabilities face many barriers to reporting abuse.[49, 56, 57] Inability to communicate what happened, information deficits, fear of the abuser, and concern about getting into trouble can complicate the disclosure. For example, if a patient reports they were "raped," eliciting information from their perspective on what happened may be important. For example, "What do you think rape is?" or "Tell me more about what your partner did to you." When people with Down syndrome do disclose, they are less likely to be believed. Struggles with expressive language, lack of sexuality education, and difficulty with time concepts can lead to frustration for families and investigators.[5, 58] If disclosure happens, remind the patient it is not their fault and praise them for being brave enough to tell you what happened.

Laws and regulations regarding the disclosure of illegal sexual behaviors experienced by people with intellectual disabilities can vary from state to state. Providers need to be aware of the reporting rules for the state in which they practice. Consideration about mandated reporting status should be taken into account according to state laws. Generally, contacting your local Adult Protective Services or the police to report exploitation is advised. If the patient lives in a facility outside the home, your state's Disability Rights Organization should also be contacted.

CASE 7

Tamika is a 29-year-old with Down syndrome who has been in a relationship with her partner for two years. She presents with her parent, who serves as her legal guardian and is concerned about the potential for unintended pregnancy.

GUARDIANSHIP AND SEXUAL ACTIVITY

Guardianship options involving people who have intellectual disabilities have changed considerably in the last few decades and state laws supporting individuals in the area of sexuality are highly variable. In general, having a guardian does not prohibit individuals with Down syndrome from participating in sexuality education, interpersonal relationships, or consensual affection or sexual expression.[59] Many state laws recognize an individual's ability to consent is not static but instead impacted by opportunities to acquire information and engage in meaningful connections with others.[60] There are a variety of guardianship statuses with some variation between the states. Regardless of guardianship status, education regarding rights and responsibilities of sexual expression should be encouraged. Modified sexuality education and visual supports for teaching about consent and rights within dating relationships are available (see resources).[61-63]

ASSESSING FOR CONSENT

In most states, having an intellectual disability does not preclude individuals from being sexually active; however, people with intellectual or developmental disabilities are typically included within the vulnerable populations category. Components of consent should be explored to ensure informed decisions are possible. Two key aspects of consent include:

- *Information:* Does the patient have the information needed to make an informed decision about sexual activity? Because opportunities for sexuality education are more limited or ineffectively adapted, people with Down syndrome may not understand the vocabulary typically used to assess consent. For example, asking "Do you know how a woman becomes pregnant?" may be more easily understood than "Do you know what sexual intercourse is?". Modified resources exist for people with Down syndrome or other intellectual disabilities to understand how pregnancy happens and ways to protect themselves from unwanted consequences of sexual activity.[63-65]

- *Voluntariness*: A lifetime of conditioned compliance can contribute to an individual's vulnerability. Remind the patient that their body is their own and they get to decide the types of touch and affection that feel safe for them. Questions like "Does your partner ever pressure you to do sexual things you don't want to do?" or "Does your partner ever ask you to do sexual things that make you feel uncomfortable?" can be ways to assess personal choice.

Both Tamika and her mother are thankful for the suggested resources to help her understand her rights within her dating relationship.

CASE 8

Nadya is a 32-year-old with Down syndrome who presents to her primary care provider's office. Nadya's older sister recently had a baby, and Nadya has decided she wants to have a baby, too. She has never been sexually active and currently does not have a significant other, although she has had some boyfriends (also with Down syndrome) in the past. Nadya's family is concerned about her ability to care for a child and wants to know if Nadya can become pregnant.

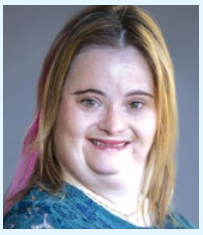
PREGNANCY

Fertility rates for women with Down syndrome are reduced, but there have been many documented cases of women with Down syndrome giving birth.[66, 67] These pregnancies appear to have few complications.[66] Most of the studies do not differentiate whether the pregnancies were intended or not. Less than half of children born to a woman with Down syndrome have Down syndrome themselves.[66] While this is not clearly understood, there may be a selection advantage for euploid (typical) as opposed to diploid (extra copy of chromosome 21) containing gametes (sperm or egg cells).[66] Women with mosaic Down syndrome are more likely to be able to become pregnant.[66]

Parenthood appears to be overall very rare for women with Down syndrome.[68] A human rights approach to individuals with intellectual disabilities parenting is an emerging topic and beyond the scope of this guide.

Nadya's family is surprised to learn that she may be fertile. They decide to talk with Nadya more about the responsibilities surrounding parenting. While she is not currently sexually active, they are concerned about her becoming unintentionally pregnant in the future and wonder if permanent sterilization is in her best interest.

SELF-ADVOCATE PERSPECTIVE: PREGNANCY



My goal is to someday be a mother and start a family. I have heard stories about pregnancy from my parents about when my mother was pregnant with me and have even searched on the web to learn more. I know that there are two different ways to give birth, cesarean or natural birth, but I know that I need to know more about pregnancy. When I am ready to have a baby, I hope my doctors can explain everything to me about what I need to know, so I know what's best for me and my future baby. I really want to be a good mom and give my little one lots of love, just like my parents have with me. With the help and support of my doctor and other health care professionals, I believe I can make my dream of having a family come true.

–Kayla M.

STERILIZATION

The topic of sterilization for women with intellectual disabilities carries many ethical questions and complications. Additionally, it is impacted by a history of questionable practices. In the 20th century, women with intellectual disabilities from a variety of countries were sterilized without their consent.[69] While the many ethical questions and implications of sterilization of women with Down syndrome are beyond the scope of this guide, it may come up in conversation with women with Down syndrome and their families. Discussing why a family is interested in sterilization may be helpful, as some families are interested in sterilization to prevent pregnancy in case of sexual abuse. However, sterilization does not prevent sexual abuse, and working with families to address their core concern is an important part of the discussion. There are legal and ethical nuances around sterilization. Using the resources of institutional ethics panels can also be helpful when navigating this topic, as there may be differences in various country and state laws.

CASE 9

Heather is a 46-year-old with Down syndrome who presents to her primary care provider's office with cessation of her period. Heather has not had her period for the past year. She is otherwise in good health and participating in her usual activities. She and her family wonder if she is in menopause and if she has any specialized bone health risks associated with menopause.

MENOPAUSE

Menopause, the normal cessation of periods as a woman ages, appears to occur at a younger age in women with Down syndrome.[70-73] The estimated average age of menopause in women with Down syndrome is between ages 44 and 46.[72, 73] There is speculation that an earlier age of menopause in women with Down syndrome may be a result of premature aging, although this has not been proven.[73]

Women with Down syndrome may experience some of the same common symptoms of menopause as women without Down syndrome including hot flashes, trouble with sleep, vaginal dryness, changes in mood, and loss of bladder control. It may be challenging for some women with Down syndrome to express and explain some of the symptoms they are experiencing, so these symptoms may not be reported. Hot flashes are a particularly difficult concept for many women with Down syndrome to describe. Sometimes, caretakers of women with Down syndrome may notice and report symptoms of menopause before the woman with Down syndrome.

Some women with Down syndrome report confusion and memory difficulties as they experience menopause. It can be difficult to determine if these symptoms are related to menopause or the beginning of Alzheimer's disease. Women with Down syndrome have a relatively short interval between the age of menopause and the potential onset of dementia.[74] One study found a significant correlation between age at menopause and diagnosis of dementia in women with Down syndrome, with early age at menopause being associated with a 1.8-fold increased risk of dementia.[71] The same study also found a correlation

between age of menopause and mortality.[71] There has been some interest in the association between estrogen and dementia risk in women with Down syndrome. Studies have shown early age of menopause and subsequently low estrogen levels in women with Down syndrome are associated with earlier age of onset for dementia.[74] There has been some interest in investigating whether impacting estrogen levels in postmenopausal women with Down syndrome with hormone replacement therapy affects the age at onset of Alzheimer's.[74] More research is needed to understand the role of estrogen on memory in menopausal women with Down syndrome. There have been no studies to date on hormone replacement therapy in women with Down syndrome. Therefore, the treatment of menopausal symptoms in women with Down syndrome is often very similar to the treatment of menopausal symptoms in women without Down syndrome.

A visual story example is available in the provider resources section to help a woman with Down syndrome understand and express her symptoms around menopause.[75]

OSTEOPOROSIS

Osteoporosis is reportedly more common in women with Down syndrome than women without Down syndrome.[28] Bone mineral densities (as measured by DEXA scans) are usually reported to be low in women with Down syndrome, which suggests an increased risk of fracture.[76] However, despite the increased risk of fracture for women with Down syndrome (based on bone mineral density and other factors), there is little data confirming if women with Down syndrome experience higher rates of fracture.[77] Additionally, the utility of DEXA scans in diagnosing osteoporosis in women with Down syndrome is not well understood. Populations with shorter statures or smaller bodies may require volumetric bone mineral density measurements to accurately characterize fracture risk.[29] In these populations, DEXA scans may report lower bone mineral density than a volumetric bone mineral density would report. Therefore, while DEXA scans may report lower bone mineral densities in women with Down syndrome, they are not helpful for assessing risk of osteoporotic fracture in women with Down syndrome.[29]

Similarly, FRAX and other fracture risk estimation tools are derived from epidemiological data from the general population and are not specific to women with Down syndrome.[29] They have limited applications for women with Down syndrome. In the JAMA article, Medical Care of Adults with Down Syndrome: A Clinical Guideline, the authors wrote that "there is insufficient evidence to recommend for or against applying established osteoporosis screening guidelines" to adults with Down syndrome. Instead, a shared decision-making approach should be implemented to meet the needs and preferences of the individual patient.[29]

There are no studies to date assessing the treatment of osteoporosis in women with Down syndrome. There have been some studies suggesting reduced osteoblast and bone formation in people with Down syndrome as opposed to increased osteoclast or bone resorption.[78] If this is the case, it may mean that drugs (like bisphosphonates) that target these pathways may have limited utility or effectiveness in women with Down syndrome.[78] More studies assessing treatment of osteoporosis in women with Down syndrome are needed.

In women with Down syndrome who do sustain a fragility fracture, an evaluation for secondary causes of osteoporosis should be initiated. The screening for secondary causes of osteoporosis should include hyperthyroidism, celiac disease, vitamin D deficiency, hyperparathyroidism, and medications that may be associated with adverse effects on bone health.[78]

Heather and her family are glad to hear that her age at menopause is typical for women with Down syndrome. She is not describing any symptoms associated with menopause, but her family now knows to watch closely for them. Heather and her family discuss osteoporosis screening. Because they are not sure that her FRAX score or a low bone mineral density on DEXA scan would change the treatment plan, they opt to forgo screening for osteoporosis at this time. They also ask if she should undergo a mammogram.

BREAST CANCER

Breast cancer is significantly less common in women with Down syndrome.[44, 70, 79] The United States Preventive Service Task Force recommends a mammogram every other year starting at age 40. A computer model shows that if the 2016 USPSTF recommendation (starting at age 50) is followed for women with Down syndrome, it takes nearly 17,000 mammograms to save one life (compared to about 2,200 in women without Down syndrome).[80] Due to false negative mammograms, the computer model also calculated it would take over 200 biopsies to save one life. Many women with Down syndrome require anesthesia to do a biopsy, and the risks from anesthesia are greater for people with Down syndrome.[81-83] Therefore, due to lower risk of breast cancer and a higher risk of complications from screening, many women with Down syndrome and their families choose not to undergo breast cancer screening with mammograms. While some researchers propose not screening women with Down syndrome for breast cancer, there are no formal recommendations for breast cancer screening in women with Down syndrome.[45] Notably, there are no separate data regarding the risk of breast cancer and screening for breast cancer in women with Down syndrome who also have a family history of breast cancer.

SELF-ADVOCATE PERSPECTIVE: OSTEOPOROSIS

People with Down syndrome can get osteoporosis at an early age. As a Special Olympics athlete and someone who comes from a sports family, I want to do everything I can to avoid getting osteoporosis so I can continue to live an active lifestyle and be like my sisters.

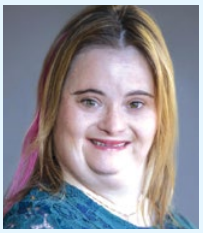
Every year, I see my doctor for a wellness and sport physical where she takes x-rays and tests my calcium and thyroid levels to make sure they are at healthy levels to prevent osteoporosis. Because it is so important to me to keep my bones healthy, I work on muscle toning twice a week to help support my bones and spend time every week learning about eating and preparing healthy foods online with friends and cooking with my mother.

Doctors can help women with Down syndrome improve or reverse the effect of osteoporosis if it is caught early. Doctors play an important role in teaching patients how to take care of themselves, so they can live fulfilling, active lifestyles.

–Ashley J.

Heather's provider discusses the low risk of breast cancer in women with Down syndrome with Heather and her family. Because Heather has a family history of breast cancer, she and her family choose to pursue a mammogram for screening purposes. Her mammogram was negative for breast cancer.

SELF-ADVOCATE PERSPECTIVE: BREAST CANCER SCREENING



My doctor has encouraged me to periodically perform breast self-exams as a proactive health measure, but as I approach the age where mammograms might become a regular part of my medical check-ups, I have a lot of questions. Do I need to worry about breast cancer? How should I prepare for an exam? What is the screening process like? I have heard from others that the process isn't invasive, but when the time comes for me to actually do these tests, having my doctor guide me through the procedure step by step would help put my mind at ease. Understanding each aspect of the screening process would help me to feel more in control of my own health and more comfortable with the entire process.

–Kayla M.

CASE 10

Yuki is a 50-year-old with Down syndrome who presents to her primary care provider's office for routine care. Her sister was recently diagnosed with fibroids and abnormal uterine bleeding, and Yuki's family wonders if she is at increased risk for this because of her diagnosis of Down syndrome. They also ask about her risk for ovarian and endometrial cancer and if she needs any screening for these cancers.

ABNORMAL UTERINE BLEEDING

There are no studies evaluating abnormal uterine bleeding in women with Down syndrome. The investigation into causes of abnormal uterine bleeding in women with Down syndrome should be the same as for women without Down syndrome. Women with Down syndrome may require extra support or even anesthesia if procedures are deemed necessary.

FIBROIDS

Uterine fibroids are common, and management is usually tailored to fit the patient's unique needs. While there is a case report of a woman with Down syndrome having a large fibroid, there are no data on the frequency of fibroids in women with Down syndrome.[84] The treatment of fibroids in women with Down syndrome should be similar to women without Down syndrome, but women with Down syndrome may require extra support or even anesthesia if procedures are deemed necessary.

OVARIAN CANCER AND ENDOMETRIAL CANCER

Generally, solid tumor cancers are reported to be less common in women with Down syndrome.[28] There are very limited data on ovarian and endometrial cancers in women with Down syndrome. One available study suggests that their risk of ovarian cancer is comparable to the general population.[44] Ovarian cancer screening in average-risk women is not recommended.[85] There is a case report of endometrial cancer in a woman with Down syndrome but no studies evaluating the overall risk of endometrial cancer in women with Down syndrome.[86] Women should be evaluated for endometrial cancer if they develop symptoms.[87] More information is needed about these cancers in women with Down syndrome.

Yuki's family is glad to hear that solid tumor cancers seem to be less common in women with Down syndrome. They will help Yuki to monitor her symptoms and seek medical care if she develops any abnormal uterine bleeding or other new symptoms.

CONCLUSION

Women with Down syndrome have some unique health care needs. While there are some differences, there are also many similarities to women without Down syndrome. Most women with Down syndrome receive health care from primary care providers. With a shared decision-making approach, providers can help women with Down syndrome and their caregivers make health care decisions that are best for each individual woman. Together, they can help each unique woman with Down syndrome to live her healthiest, best life!

Each case featured in this guidebook provides insight and information to better understand the nuances involved in caring for women with Down syndrome. By dedicating time to expand your knowledge and expertise, you are not only enhancing the quality of care you provide but also making a meaningful difference in the lives of women with Down syndrome and their families and caregivers.

Thank you for your commitment to improving the health care experiences of women with Down syndrome. Your empathy and dedication are instrumental in improving their health, well-being, and overall quality of life. We hope you will continue to learn more about Down syndrome and, after reading this guide, walk away knowing that life with Down syndrome is great—and you are helping to make it even better!

RESOURCES FOR PROVIDERS

NATIONAL DOWN SYNDROME SOCIETY (NDSS)

www.ndss.org

The NDSS Health and Wellness Program promotes improved health and well-being for all individuals with Down syndrome. Through collaboration, NDSS develops tailored and accessible resources for individuals with Down syndrome, families, and caregivers across the lifespan.

Please visit our publications page for additional resources, such as *Practicing Inclusive Mental Healthcare of Individuals with Down Syndrome*, *Alzheimer's Disease and Down Syndrome: A Practical Guidebook for Caregivers*, *End-of-Life and Down Syndrome*, and *Aging and Down Syndrome: A Health & Well-Being Guidebook*.

321go!

www.ndss.org/321go

NDSS designed the *321go!* program to promote healthy lifestyle choices in physical activity, balanced nutrition, and emotional wellness among individuals with Down syndrome and their families.

ADVOCATE HEALTH CARE ADULT DOWN SYNDROME CENTER

www.adscresources.advocatehealth.com

The Adult Down Syndrome Center at Advocate Health Care has a great library of videos and pamphlets aimed at providers, caregivers, and people with Down syndrome that discuss a variety of health and wellness topics.

AMERICAN ACADEMY OF DEVELOPMENTAL MEDICINE AND DENTISTRY (AADMD)

www.aadmd.org

AADMD aims to improve the quality of healthcare for individuals with neurodevelopmental disorders and intellectual disabilities.

DOWN SYNDROME MEDICAL INTEREST GROUP (DSMIG)

www.dsmig-usa.org

DSMIG is a group of health professionals from a variety of disciplines who provide care to individuals with Down syndrome. DSMIG-USA® educates members on the best practices of care and supports the development of Down syndrome clinics.

GLOBAL DOWN SYNDROME FOUNDATION

https://www.globaldownsyndrome.org/medical-care-guidelines-for-adults/

The GLOBAL Medical Care Guidelines for Adults with Down Syndrome (GLOBAL Adult Guidelines) provide first in-kind, evidence-based medical recommendations to support clinicians in their care of adults with Down syndrome.

KRAMER DAVIS HEALTH

www.kd.health

Kramer Davis (KD) Health provides healthcare to adults and adolescents with intellectual and developmental disabilities (IDD), including medical care, dental treatment, nutrition, psychiatry, behavioral health, physical therapy, occupational therapy, and speech pathology.

MENTAL WELLNESS IN ADULTS WITH DOWN SYNDROME: A GUIDE TO EMOTIONAL AND BEHAVIORAL STRENGTHS AND CHALLENGES (2nd Edition) McGuire, D., & Chicoine, B. (2021).

www.adscresources.advocatehealth.com/mental-wellness-in-adults-with-down-syndrome-2nd-edition

Mental Wellness in Adults with Down Syndrome: A Guide to Emotional and Behavioral Strengths and Challenges. This easy-to-read guide clarifies what are the common behavioral characteristics of Down syndrome, how some can be mistaken for mental illness, and what are the mental health problems that occur more commonly in people with Down syndrome. In addition, the authors discuss the importance of regular assessment and how behavior and mental well-being can be affected by environmental conditions, social opportunities, and physical health.

MENSTRUATION AND MENOPAUSE VISUAL SUPPORTS

RESOURCES FOR WOMEN WITH DOWN SYNDROME AND CAREGIVERS

Advocate Health Care Adult Down Syndrome Center Resource Library:

www.adscresources.advocatehealth.com

- Menstruation
  www.adscresources.advocatehealth.com/resources/menstruation-visuals
- Menopause
  www.adscresources.advocatehealth.com/resources/menopause-visuals

PELVIC EXAM SOCIAL STORIES

Autism Services, Education, Resources, and Training Collaborative (ASERT)
Pelvic exam social story:
www.paautism.org/resource/pelvic-exam-social-story

Hesperian Health Guides: *A Health Handbook for Women with Disabilities*. By Maxwell J, Watts Belser J, David D.
The pelvic exam:
www.en.hesperian.org/hhg/A_Health_Handbook_for_Women_with_Disabilities:The_pelvic_exam

SEX EDUCATION

Advocate Health Care Adult Down Syndrome Center Resource Library:
www.adscresources.advocatehealth.com

Boyfriends & Girlfriends: A Guide to Dating for People with Disabilities. By Terri Couwenhoven
www.terricouwenhoven.com

Sexuality for All Abilities. Mad Hatter Wellness.
www.madhatterwellness.com

Relationships and Sex Education. Down Syndrome Association (UK).
www.downs-syndrome.org.uk/about-downs-syndrome/lifes-journey/relationships-and-sex-education

Sex Ed for People with I/DD: Consent. National Council on Independent Living.
www.youtube.com/watch?v=09m9Hv0Ajto

Sex Ed for People with I/DD: Pregnancy. National Council on Independent Living.
www.youtube.com/watch?v=FGY-lQCNI5c&list=PLuEvYNNQ-dHeVhbyeJHx9s8oqsvBk621v&index=7&t=6s

What is Sex?: A Guide for People with Autism, Special Educational Needs and Disabilities. By Kate Reynolds
www.kateereynolds.com

NDSS RESOURCES

321go!
www.ndss.org/321go
NDSS designed the *321go!* program to promote healthy lifestyle choices in physical activity, balanced nutrition, and emotional wellness among individuals with Down syndrome and their families.

Aging and Down Syndrome: A Health & Well-Being Guidebook
www.ndss.org/resources/aging-and-down-syndrome-health-well-being-guidebook
This guidebook is intended for families, professionals, direct caregivers, or anyone concerned with the general welfare of an adult with Down syndrome with a specific focus on aging and health-related matters in an individual with Down syndrome, including common medical conditions, Alzheimer's and Down syndrome, and mental health.

Alzheimer's Disease & Down Syndrome: A Practical Guidebook for Caregivers
www.ndss.org/resources/alzheimers-disease-down-syndrome-practical-guidebook-caregivers
This guidebook is specifically for those caring for an individual with Down syndrome diagnosed with Alzheimer's disease.

Self-Advocacy at Medical Appointments
www.ndss.org/resources/self-advocacy-medical-appointments
To help prepare individuals with Down syndrome to advocate for themselves at health care appointments, NDSS has created a printable two-sided worksheet that can be filled out at home prior to the appointment. Individuals should bring this completed worksheet to health care appointments to make communication with medical professionals easier and more effective.

Story About How to Change My Pad

This is how I will change my sanitary pad.

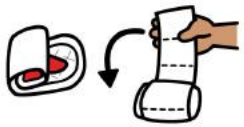
I will go to the bathroom for privacy.

I can keep clean pads and underwear in a bag so I can bring the supplies to the bathroom with me.

When I go to the bathroom, I will pull down my pants and my underwear. I will sit on the toilet

I will take the dirty pad out of my underwear.

I will fold it in half so I do NOT see the blood. Then I will wrap it in toilet paper.

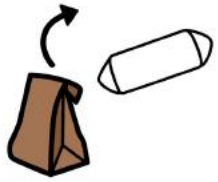


I will throw the pad and toilet paper in the garbage can.

I will NOT throw it into the toilet.

I will make sure there is no blood on my underwear. If there is, I will need to put on clean underwear.

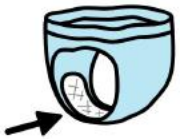
I will get a new, clean sanitary pad out of my bag.



I will open the wrapper and take the pad out.

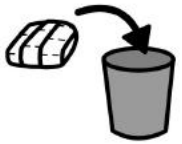
I will remove the paper so I can see the sticky tape.

I will put the clean pad in my underwear with the sticky side touching my underwear.



I will make sure the pad sticks to my underwear.

I can throw the paper away in the garbage can.



I should also use the toilet and wipe to make sure I am clean.

I will pull up my underwear and pants.

I will take my bag of supplies with me when I leave the bathroom.

I will wash my hands.

If I have questions or need help, I can ask a parent.

www.adscresources.advocatehealth.com/resources/menstruation-visuals/

Advocate Medical Group
Adult Down Syndrome Center

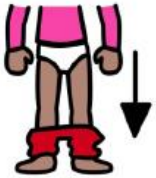
Story About How to Change My Period Panties

This is how I will change my period panties.

I will go to the bathroom for privacy.

I can keep clean period panties in a bag so I can bring the supplies to the bathroom with me.

When I go to the bathroom, I will pull down my pants and my underwear.

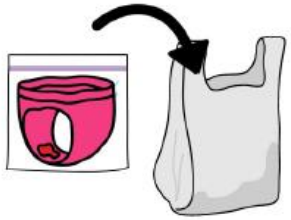


I will sit on the toilet.

I will take off my pants and my dirty period panties.

I will put my dirty period panties in the small bag.

I will put the small bag inside the larger bag.



I will take out the new pair of clean period panties and put them on with my pants.

I should also use the toilet and wipe to make sure I am clean.

I will stand up and pull up my clean period panties and pants.

I will flush the toilet.

I will take the bag of supplies with me.

I will wash my hands.

If I have questions or need help, I can ask a parent.

www.adscresources.advocatehealth.com/resources/menstruation-visuals/

Advocate Medical Group
Adult Down Syndrome Center

MENOPAUSE

www.adscresources.advocatehealth.com/resources/menopause-visuals/

Advocate Medical Group
Adult Down Syndrome Center

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