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# The Importance of Unique Healthcare Guidelines for Adults with Down Syndrome

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## Introduction

The development and implementation of healthcare guidelines is a complicated process fraught with challenges. Many factors must be considered.<sup>1-4</sup> When developing such guidelines, such as the [GLOBAL Medical Care Guidelines for Adults with Down Syndrome](#), reviewing risks and benefits is a key. The purpose of this article is to discuss specific healthcare guidelines for adults with Down syndrome, adapted from general preventative healthcare guidelines, considering three key issues.

## What Phenotypic Features of Down Syndrome Affect Medical Screening and Treatment?

### Altered Prevalence of Certain Conditions

As discussed in multiple articles in this reference, there are many characteristics associated with trisomy 21 that impact an individual's health, ability to participate in medical care, response to treatment, and other factors. Although there are limited data on how these features impact the risks and benefits of specific screening tests, one recent study gives some insight.<sup>5</sup> Because there is a lower prevalence of breast cancer in women with Down syndrome,<sup>6,7</sup> researchers determined that many more mammograms were required to

save one life from breast cancer in women with Down syndrome compared to women without Down syndrome. In addition, many more biopsies were required for women with Down syndrome with false positive mammograms, with the potential for anesthesia complications as noted below. The lower risk of breast cancer and the greater risks associated with screening and diagnosis indicate a need to develop a Down syndrome specific evidence-based mammography screening guideline.

### Life Expectancy

In the United States, the [life expectancy](#) of individuals with Down syndrome is about 60 years of age in the white population, and lower in other races. This is significantly lower than the United States' average life expectancy of 77.5 years (2022 data).<sup>8</sup> Life expectancy impacts recommendations for screening measures because those with limited life expectancy have fewer potential years of life to gain, have more comorbid conditions,<sup>9</sup> and may experience more complications from testing and treatment.<sup>10</sup> For this reason, the U.S. Preventive Services Task Force sets upper age limits for many recommended screenings.<sup>11</sup>

### Family History

Little data are available regarding the impact of family genetics on health issues for individuals with Down syndrome. It is mostly unclear how the genetics of Down syndrome interacts with family genetics, although it is assumed that family genetics may have some impact.

## Quality of Life Goals

Finally, some individuals with Down syndrome may feel that medical interventions to screen and treat for some illnesses are too onerous to be worth the possible benefits. As for all medical interventions, these decisions require careful shared decision-making.

## What Characteristics of the Testing and Treatment Affect the Benefits and Risks?

Perhaps the biggest challenge in crafting healthcare guidelines for adults with Down syndrome is the lack of population-specific data. For any given test, the false positive and false negative rates, positive and negative predictive values, or other data normally used to evaluate the risks and benefits to the patient are unknown. This limits testing and treatment recommendations.

The “cost” of screening includes both economic<sup>12</sup> costs as well as the clinical risks associated with participating in the screening. When considering the financial costs, there are potential ethical issues with regards to limiting care for adults with Down syndrome. Discussing fiscal cost is beyond the scope of this article and has not been studied in individuals with Down syndrome.

An important clinical risk consideration is that many individuals with Down syndrome will have

challenges participating in the initial screening test, the follow up testing, or the indicated treatment. For example, an individual with Down syndrome may have difficulty communicating with the care team or understanding the potential benefits of testing procedures. Sensory issues, co-occurring conditions (e.g., autism), or previous negative healthcare experiences are also common. Nevertheless, testing and treatment may be acceptable to the individual and successfully completed with appropriate behavioral supports, communication aids, and other assistance as addressed in the article [Supporting an Adult with Down Syndrome in the Healthcare Setting.](#)

If those techniques are not successful, limited cooperation may necessitate the use of sedation or anesthesia to complete testing and treatment in some individuals with Down syndrome. This includes procedures for which sedation or anesthesia are not required by most individuals without an intellectual disability. Due to a variety of physiological, anatomical, and co-occurring conditions, there is potential for increased anesthesia complications in individuals with Down syndrome.<sup>13, 14</sup> There are limited data regarding how the need for anesthesia in testing affects the overall outcome of health screening in adults with Down syndrome.

## What Characteristics of the Condition Affect Screening and Treatment?

As noted above, the lack of data in individuals with Down syndrome limits our ability to provide guidance in answering this question for many conditions. If there is a pre-clinical or early clinical phase of the condition in which early detection and treatment are demonstrated to positively impact outcomes, then testing may be warranted.

## Healthcare Guidelines for Adults with Down Syndrome

GLOBAL Medical Care Guidelines for Adults with Down Syndrome were published in 2020.<sup>15</sup>

Note that these recommendations vary in emphasis (weak to strong) based on the quality of evidence (low to moderate). Nevertheless, they provide the first evidence-based guidelines for adults with Down syndrome in the United States. These guidelines are presently undergoing review and may be expanded to include new recommendations and additional conditions.

Some highlights include:

- Screen for diabetes mellitus every 2-3 years starting at age 21 if also obese, or at age 30 if not obese.

- For those with a history of congenital heart disease, provide regular follow-up with a cardiologist throughout adulthood.
- Assess annually for signs and symptoms of cervical myelopathy associated with atlantoaxial instability (AAI). Routine neck X-rays are not recommended. More information on AAI is available in the article [Common Health Conditions in Adults with Down Syndrome](#).
- Assess annually for gastrointestinal and non-gastrointestinal signs and symptoms of celiac disease and follow up with blood testing as needed. Additional information is available in the article [Common Health Conditions in Adults with Down Syndrome](#).
- Order blood testing to assess for hypothyroidism every 1-2 years. Additional information is available in the article [Common Health Conditions in Adults with Down Syndrome](#).
- Order a lipid profile every 5 years starting at age 40 and use a 10-year risk calculator to determine need for a statin.
- For any adult with Down syndrome presenting with a mental health disorder, assess for contributing physical conditions. For more information, see the articles [Mental Health: Diagnosis and Treatment of Adults with Down Syndrome](#) and [Decline in Skills and Behavioral Change in Adults with Down Syndrome](#).

- Begin annual screening for Alzheimer's at age 40. Be cautious when diagnosing Alzheimer's disease in individuals with Down syndrome who are less than 40 years old, due to low prevalence of symptomatic Alzheimer's disease before that age. More information is available in the article [Alzheimer's Disease in Adults with Down Syndrome](#).

More information about these conditions is available in the [Common Health Conditions in Adults with Down Syndrome](#) article.

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