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Is a High Index of Clinical Suspicion Required for Turner Syn-drome Mosaicism after a Diagnosis of Coeliac Disease

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Introduction

Turner syndrome mosaicism and coeliac disease are both distinct medical conditions, but their co-occurrence in a patient can present unique challenges, Turner syndrome, a chromosomal disorder affecting females, results from the complete or partial absence of one of the X chromosomes, while coeliac disease is an autoimmune disorder triggered by gluten consumption, leading to damage in the small intestine. When these conditions occur together, it requires a high index of clinical suspicion to recognize the potential overlap, especially as the symptoms of one condition may mask or complicate the diagnosis of the other [1]. Turner syndrome, while typically associated with short stature, infertility, and a range of other physical abnormalities, is not always easy to diagnose, particularly in its mosaic form. Turner syndrome mosaicism refers to a situation where some of the cells in a person's body have the normal two X chromosomes, while others have only one X chromosome, leading to a less severe presentation of the syndrome compared to the classic form. The mosaic nature of the condition means that symptoms can vary greatly, and in some cases, the features may be subtle enough that a diagnosis is not immediately obvious [2].

The clinical presentation of Turner syndrome can vary widely. Some individuals with Turner syndrome may have very few physical signs and symptoms, while others may exhibit a range of conditions such as heart defects, kidney abnormalities, hearing loss, or delayed puberty. Because of the variability in symptoms, Turner syndrome is often not diagnosed until a patient is evaluated for other unrelated health issues, which can complicate the detection of the disorder. In fact, many girls with Turner syndrome, particularly those with mosaic forms, may not be diagnosed until later in life when infertility issues or other health problems arise. It is therefore crucial for clinicians to have a high level of clinical suspicion when dealing with patients presenting with atypical or unexplained symptoms, particularly when those symptoms involve growth or development. Coeliac disease, on the other hand, is an autoimmune disorder where the ingestion of gluten triggers an immune response that damages the lining of the small intestine. The disease is often associated with gastrointestinal symptoms like diarrhea, abdominal pain, and bloating, but it can also manifest with a range of non-gastrointestinal symptoms, such as skin rashes, fatigue, and infertility. Coeliac disease is diagnosed through a combination of blood tests (looking for antibodies against gluten) and a biopsy of the small intestine to assess the extent of the damage.

Description

Both Turner syndrome and coeliac disease can present with gastrointestinal symptoms, such as abdominal discomfort, bloating, and growth issues. These overlapping symptoms can sometimes delay a correct diagnosis, as they may be attributed solely to one condition, ignoring the possibility of the other. Additionally, patients with Turner syndrome may experience growth failure, which can be further exacerbated by the malabsorption associated with coeliac disease. This can make it difficult to determine whether a patient's growth concerns are primarily due to Turner syndrome or the underlying effects of undiagnosed coeliac disease. The interplay between the two conditions is important to understand in order to guide clinicians in the diagnostic process. Coeliac disease has been noted to occur more frequently in individuals with Turner syndrome than in the general population. This elevated risk suggests that coeliac disease should be considered as part of the differential diagnosis in any patient with Turner syndrome, especially if they present with gastrointestinal symptoms or unexplained growth issues. However, the reverse is also true: individuals with coeliac disease may have subtle signs of Turner syndrome, which could go unnoticed unless a thorough evaluation is undertaken [3].

Because both Turner syndrome and coeliac disease can be asymptomatic in their early stages or present with vague symptoms, clinicians need to maintain a high index of suspicion. Turner syndrome may be missed if a patient does not present with the more recognizable features, such as short stature or a webbed neck, while coeliac disease may not be diagnosed if the patient does not exhibit obvious gastrointestinal symptoms. As both conditions can also present with infertility, patients who are evaluated for infertility issues may benefit from screening for both Turner syndrome and coeliac disease, particularly if they have risk factors for either condition [4].

The diagnosis of Turner syndrome mosaicism after the diagnosis of coeliac disease requires careful clinical evaluation and attention to detail. While coeliac disease may lead to nutritional deficiencies that can affect growth and development, the underlying chromosomal abnormality in Turner syndrome can contribute to a variety of health problems that might not be immediately linked to coeliac disease. An accurate diagnosis requires a clinician to consider the full spectrum of symptoms and risk factors, including a patient's medical history, family history, and any previous treatments or diagnoses. In some cases, a karyotype analysis or genetic testing may be necessary to confirm the diagnosis of Turner syndrome mosaicism, particularly if a patient presents with atypical features that do not clearly align with the classic presentation of either condition. For individuals who are diagnosed with both Turner syndrome and coeliac disease, management becomes more complex. Treatment for coeliac disease typically involves strict adherence to a gluten-free diet, which can improve gastrointestinal symptoms and prevent further damage to the small intestine. However, for patients with Turner syndrome, additional treatments may be required to address specific health issues, such as hormone replacement therapy for puberty or growth hormone therapy for short stature. Monitoring and managing both conditions concurrently require a multidisciplinary approach, with input from specialists in genetics, gastroenterology, endocrinology, and other relevant fields [5].

Conclusion

The co-occurrence of Turner syndrome mosaicism and coeliac disease necessitates a high index of clinical suspicion, as the overlapping symptoms of both conditions can complicate the diagnostic process. While each disorder has its own distinct features, their concurrent presence may result in a more

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complex clinical picture. Clinicians must be aware of the potential for both conditions to occur together and should consider a thorough evaluation, including genetic testing and screening for coeliac disease, when presented with patients exhibiting relevant symptoms. A timely and accurate diagnosis is essential to ensure appropriate treatment and improve patient outcomes, as both conditions require ongoing management and monitoring to address their unique challenges.

Acknowledgement

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Conflict of Interest

None.

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