Early Bone Loss in Turner Syndrome: A Case of Primary Ovarian Insufficiency and Osteoporosis

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Introduction

- TS is a rare chromosomal disorder characterized by the partial or complete loss of one X chromosome, leading to multiple comorbidities, including ovarian insufficiency and osteoporosis [1,2] (Figure 1).
- These patients are at a higher risk for fractures even with normal bone mineral density (BMD) due to estrogen deficiency [3] (Figure 2).
- While hormone replacement therapy (HRT) is widely recommended to prevent bone loss, limited data exists on secondary fracture prevention in young TS patients with established osteoporosis [4].

Objectives

This case highlights the challenges in managing osteoporosis in a young patient with Turner syndrome (TS) who developed the condition prematurely. Given the lack of established guidelines for this specific patient population, this case underscores the need to explore treatment strategies tailored to address early-onset osteoporosis in TS patients.

Case Description

- A 23 year old Hispanic female with TS, primary amenorrhea, and secondary hypertension who presented to establish primary care. Diagnosed with TS at age 5, she experienced delayed initiation of growth hormone therapy and experienced inconsistent follow-up due to a lack of insurance and low health literacy.
- **Physical Exam**
  - Vitals: BP 160/29 mmHg, otherwise within normal limits.
  - Short stature, broad chest with widely spaced nipples, and webbed neck.
- **Labs**
  - Vitamin D levels 7.5 mg/mL, indicating deficiency.
  - Estradiol levels were undetectable (<5), reflecting poor adherence to HRT.
  - Bone density scan confirmed early-onset osteoporosis with significantly low z-scores across the lumbar spine (-3.6), femoral neck (-2.5), and total hip (-2.7) (Table 1).
- **Clinical Course**
  - The patient was started on vitamin D supplementation, estradiol 1mg and micronized progesterone 200mg once daily during the first 12 days of the month.
  - After consultation with endocrinology, secondary causes of osteoporosis were ruled out and Alendronate 70 mg weekly was initiated to mitigate fracture risk.
  - Follow-up appointments were scheduled to monitor therapy adherence and BMD progress with yearly bone density scan.

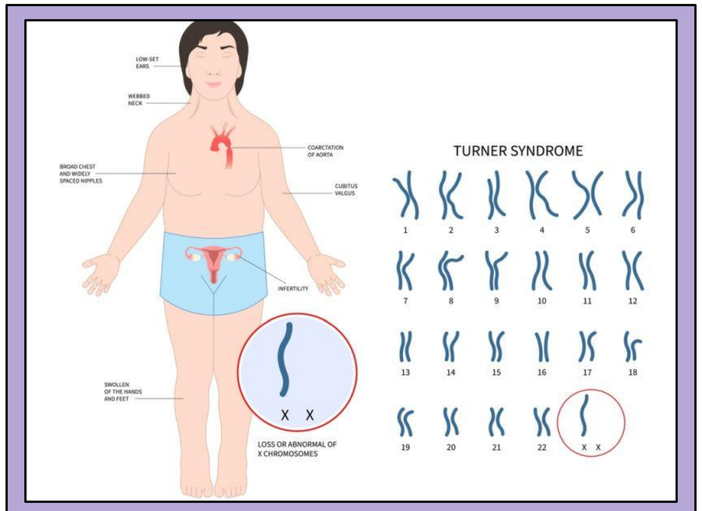

Figure 1: Phenotypic characteristics and genetic makeup of Turner Syndrome.

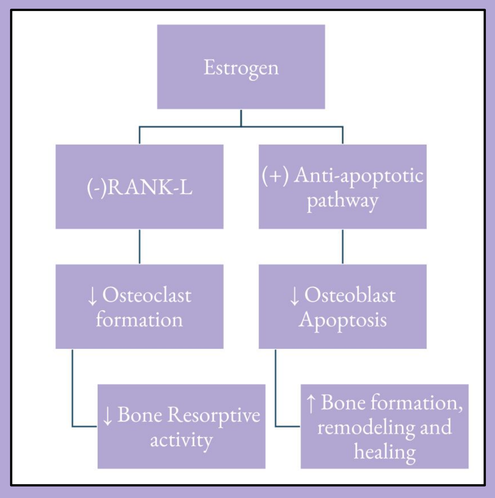

Figure 2: Estrogen effects on bone homeostasis, (-): inhibits, (+) promotes, (↑): increases (↓): decreases

Parameter	Lumbar Spine (L1-L4)	Left Femoral Neck	Left Total Hip
BMD*	0.753 g/cm2	0.696 g/cm2	0.679 g/cm2
T-score	-3.6	-2.5	-2.6
Z-score	-3.6	-2.5	-2.7

Table 1: Patient's Bone Density Scan test results.
*BMD: bone mass density

Discussion

- The management of osteoporosis in TS patients is complex, requiring a combination of pharmacological and lifestyle interventions.
- Hormone replacement and vitamin D optimization are critical while assessing the effectiveness of bisphosphonate therapy in improving bone health for patients that have TS.
- OMT techniques such as functional and soft tissue approaches may enhance joint mobility, proprioception, and postural stability, reducing fracture risk and improving quality of life [10].
- By addressing hyperkyphosis and muscle weakness, OMT could complement medical treatments to optimize outcomes [11].
- This case also highlights the critical role of addressing social determinants of health, such as insurance access and health literacy, which can significantly impact adherence to preventative care.

Conclusion

- A multidisciplinary approach incorporating endocrinology consultation, tailored HRT, bisphosphonate therapy, and potentially OMT will result in improved adherence and stabilization of BMD in this patient.
- This case highlights the importance of addressing patient-specific barriers, including health literacy and access to care, while integrating innovative treatments like OMT.
- Future follow-up will be crucial to evaluate the long-term effectiveness of these interventions in improving health outcomes for young TS patients with early-onset osteoporosis.

Acknowledgements

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References

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