# Look me in the Eyes Presented by Jessica Carmel PGY3 - Internal medecine Sherbrooke University

## **Evidence of Conflict of Financial Interest**

	Co-author	Conflict disclosures
1.	Jessica Carmel	None

## Look me in the Eyes

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### Introduction

Severe hip pain during pregnancy is often attributed benian musculoskeletal to changes. 1 However, in rare circumstances, it may signal an underlying pathology with potentially devastating consequences for both mobility and quality of life. When standard investigations are inconclusive and symptoms continue to worsen, clinicians are faced with a challenging diagnostic puzzle. We present the case of a young woman in her third trimester who developed progressive bilateral hip pain, ultimately revealing an unexpected and multifactorial etiology.

### Learning objectives

- 1. Identify the various causes of fractures during pregnancy.
- 2. Recognize the clinical signs indicative of osteogenesis imperfecta.

### Case

A 30-year-old woman was referred for evaluation of worsening hip pain over several weeks, for which she had already sought medical attention multiple times. She was 34 weeks into her first pregnancy.

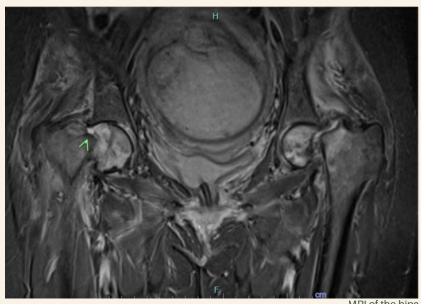
Aside from two fractures of the ankles and one metacarpal due to low-impact trauma in childhood, she is in good health with no relevant family history.

She has been experiencing progressive pain in both hips since the second trimester, eventually leading to difficulty with mobility and then a fall that left her unable to walk.

On examination, she appeared short-statured and underweight, and was in severe pain. No active movement of the proximal lower limbs was possible, and distal strength was reduced to 2/5 with an antalgic pattern. Even minimal passive movement caused unbearable pain. Sensation and deep tendon reflexes were preserved.

Initial blood tests revealed unremarkable blood count, CK, TSH, and CRP levels.

After consultation with the neurology and rheumatology teams, transient osteoporosis of the hips was considered the most likely diagnosis. Inflammatory and neurological involvement of the lower limbs seemed less likely based on the physical exam and work-up. Given the pregnancy context, an MRI of the hips were ordered.



MRI of the hips

displaced Imaging revealed bilateral transcervical fractures with avascular necrosis of the femoral heads and bone marrow edema.

Further questioning revealed that the patient had a very low calcium intake before and during pregnancy. Her calcium-phosphate panel showed normal calcium, phosphorus, and vitamin D levels, but slightly decreased PTH.

One detail eventually caught a colleague's attention: the patient had abnormally blue sclerae.



Patient's eyes

### Discussion

Hip pain in pregnancy is relatively common and is often attributed to musculoskeletal pelvic girdle pain, or transient strain, osteoporosis of the hip (TOH).<sup>1</sup>

TOH is a rare condition, typically presenting in the third trimester, characterized by acuteonset hip pain, limited mobility, and reversible bone marrow edema visible on MRI.<sup>2</sup> In a quarter of affected women, this condition can be complicated by a fracture.<sup>3</sup>

Osteonecrosis of the femoral head results from vascular compromise either traumatic or atraumatic and is characterized by hip pain, worsened by weight-bearing and progressively leading to reduced range of motion.4 During pregnancy, this is extremely rare and of uncertain etiology. Diagnosis is established with CT, or MRI.5

However, the presence of multiple lowimpact fractures in childhood and the physical finding of blue sclerae pointed toward an alternative etiology. Osteogenesis imperfecta (OI) type 1, the mildest and most common form, frequently goes undiagnosed until adulthood, as affected individuals may only present with subtle features such as short stature, hearing loss, or recurrent fractures. In this patient, the diagnosis was delayed until pregnancy unmasked the severity of her skeletal fragility. 6

In OI, mutations affecting type I collagen synthesis lead to reduced bone strength and increased fracture risk.<sup>6</sup> These collagen damages reduces the thickness of the sclera, making it more translucent and allowing the underlying color of the choroid, rich in blood vessels, to show through, which explains the blue sclera.<sup>7</sup>

Pregnancy itself further challenges maternal bone homeostasis, as calcium demand rises significantly during the third trimester to support fetal skeletal development.<sup>8</sup>

Although intestinal calcium absorption normally increases, women with insufficient calcium intake, low bone mass, or pre-existing skeletal disorders may experience accelerated bone loss. Bone fragility persists during breastfeeding, since bone resorption is the main mechanism regulating the calcium requirements needed for lactation.8

### Conclusion

We concluded that the etiology of the fractures is likely multifactorial.

We strongly suspect they are secondary to bone fragility due to type 1 osteogenesis imperfecta (which was later confirm by the genetic testing), pregnancy-induced osteoporosis, and possibly transient osteoporosis of the hips. We believe the avascular necrosis is secondary to the fractures.

As the patient remained in severe pain despite analgesia, a cesarean was performed at 35 weeks, followed by bilateral total hip replacement with prophylactic cerclage. She was prescribed calcium and vitamin D supplements and was strongly advised not to breastfeed.<sup>9</sup>

This case highlights several important teaching points. First, not all hip pain in pregnancy is benign; persistent or severe symptoms should prompt timely imaging, preferably MRI, to rule out fractures or bone pathology. Second, clinicians should consider OI in adults with a history of multiple childhood fractures, even in the absence of a known family history.

### References

- 1. Bermas, B. L. Maternal adaptations to pregnancy: Musculoskeletal changes and pain. UpToDate, Connor RF (Ed), Wolters Kluwer.
- 2. Asadipooya, K., Graves, L., & Greene, L. W. (2017). Transient osteoporosis of the hip: review of the literature. Osteoporosis international: a journal established as result of cooperation between the European Foundation for Osteoporosis and the National Osteoporosis Foundation of the USA, 28(6), 1805–1816. <a href="https://doi.org/10.1007/s00198-017-3952-0">https://doi.org/10.1007/s00198-017-3952-0</a>
- 3. Morton, A., & Savard-Heppel, J. (2024). Transient osteoporosis of the hip in pregnancy. Obstetric medicine, 17(4), 188–193. https://doi.org/10.1177/1753495X241236222
- 4. Baig, S. A., & Baig, M. N. (2018). Osteonecrosis of the Femoral Head: Etiology, Investigations, and Management. *Cureus*, 10(8), e3171. <a href="https://doi.org/10.7759/cureus.3171">https://doi.org/10.7759/cureus.3171</a>
- 5. Montella, B. J., Nunley, J. A., & Urbaniak, J. R. (1999). Osteonecrosis of the femoral head associated with pregnancy. A preliminary report. The Journal of bone and joint surgery. American volume, 81(6), 790–798. https://doi.org/10.2106/00004623-199906000-00006
- 6. Balasubramanian, M. Oeteogenesis imperfecta: An Overview. UpToDate, Connor RF (Ed), Wolters Kluwer.
- 7. Treurniet, S., Burger, P., Ghyczy, E. A. E., Verbraak, F. D., Curro-Tafili, K. R., Micha, D., Bravenboer, N., Ralston, S. H., de Vries, R., Moll, A. C., & Eekhoff, E. M. W. (2022). Ocular characteristics and complications in patients with osteogenesis imperfecta: a systematic review. Acta ophthalmologica, 100(1), e16–e28. https://doi.org/10.1111/aos.14882
- 8. Kovacs CS. Maternal Mineral and Bone Metabolism During Pregnancy, Lactation, and Post-Weaning Recovery. Physiol Rev. 2016 Apr;96(2):449-547. doi: 10.1152/physrev.00027.2015. PMID: 26887676.
- 9. Eugénie Koumakis, Valérie Cormier-Daire, Azeddine Dellal, et al. Osteogenesis Imperfecta: characterization of fractures during pregnancy and post-partum. *Orphanet Journal of Rare Diseases*. 2022;17(1):1-12. doi:10.1186/s13023-021-02148-x