Paget’s Disease of the Bone

Question 1

Correct Answer: C

Discussion
The best answer is C because asymptomatic patient but with lesions in the skull, spine, weightbearing bones, or abutting joints would benefit from pharmacotherapy to promote mobility. A could be correct if it were a symptomatic patient with and elevated alkaline phosphatase. B could be correct if it were a symptomatic patient and imaging consistent with active Paget’s disease. D could be correct if it were an asymptomatic patient with alkaline phosphatase levels 2 times the upper limit of normal.

Osteogenesis Imperfecta

Question 1

Correct Answer: A

Discussion
The best answer is A. B describes McCune-Albright syndrome and C describes hereditary hypophosphatemic rickets.

Question 2

Correct Answer: A

Discussion
The best answer is A because autosomal dominant mutations in COL1A1 or COL1A2 account for 90% of Osteogenesis Imperfecta; furthermore, males and females are equally affected.

Question 3

Correct Answer: C

Discussion
The best answer is C because patients with IFITM5 mutations develop hyperplastic calluses after trauma.

Question 4

Correct Answer: B

Discussion
The best choice is B because this is likely OI type 2 because the newborn died. A unlikely because the
patient likely does not have OI type 1, the mildest form. C is unlikely because OI types from recessive mutations occur in less than 5% of cases.

McCune-Albright Syndrome

**Question 1**

*Correct Answer: A*

**Discussion**
The best answer is A: Malignant transformation is possible so avoid I-131; Tx hyperthyroidism with MMI or surgery.

Hypophosphatemia

**Question 1**

*Correct Answer: C*

**Discussion**
The best answer is C because FGF23 causes phosphate wasting from the serum into the urine for hereditary hypophosphatemic rickets.

**Question 2**

*Correct Answer: C*

**Discussion**
The best answer is C because the labs typically show a low serum phosphorus, high urine phosphorus, high or normal PTH, and high FGF23.

**Question 3**

*Correct Answer: C*

**Discussion**
The best answer is C because 80 to 90% of hereditary hypophosphatemic rickets are X-linked dominant. The other patterns are possible but less common. This is in contrast to OI where 90% of cases are autosomal dominant.

**Question 4**

*Correct Answer: C*
Discussion
The best answer is C. A leads to OI, B leads to MAS, and D leads to autosomal recessive hypophosphatemic rickets – not the X-linked form.

Question 5
Correct Answer: A

Discussion
The best answer is A. B and C are possible second-line treatments. D can help OI, OP, or MAS.

Question 6
Correct Answer: D

Discussion
The best answer is D because patients want strong bones; it is possible to see epiphyseal healing on x-rays. A is incorrect because it would promote nephrocalcinosis and secondary hyperparathyroidism. B is incorrect because patient can actually experience catch-up growth with therapy. C is incorrect because the aim to is to normalize Alk phos. E is incorrect because this can also promote nephrocalcinosis.

Hypophosphatasia

Question 1
Correct Answer: D

Discussion
D is the best answer. COL1A1 mutations cause Osteogenesis imperfect. GNAS mutations cause Albright's hereditary osteodystrophy and McCune Albright syndrome. PHEX mutations cause Hypophosphatemia.

Question 2
Correct Answer: C

Discussion
The best answer is C because patients will have high pyridoxal 5'-phosphate levels. Alkaline phosphatase will actually be low, inorganic pyrophosphate will be high, and Phosphoethanolamine will also be high.
Question 3
Correct Answer: A

Discussion
The best answer is A because the other choices have had limited success.

Albright Hereditary Osteodystrophy

Question 1
Correct Answer: D

Discussion
The best answer is D because there is no PTH resistance by the labs. If there had been PTH resistance, the best answer would have been C. A causes hypoparathyroidism and B causes familial hypocalciuric hypercalcaemia.

Question 2
Correct Answer: B

Discussion
The best answer is B because it is ok to target normal serum calcium levels, low-normal serum calcium goals are not required like they are in hypoparathyroidism. A is not a good choice because immobilization can cause joint stiffness. C is incorrect, patients generally require less food because their resting energy expenditure is lower than BMI-matched controls. D is incorrect, PTH analogs are not helpful because of PTH resistance.