MLS Nomenclature Practice

1. 47,XY,+13
   Male with trisomy 13

2. 45,XX,der(14;21)(q10;q10)
   Normal female with Robertsonian between 14 and 21 **NOTE:** This is a 45 count because the 14 and 21 are together

3. 46,XX,der(14;21)(q10;q10),+21
   Female with Robertsonian between 14 and 21and with 2 normal 21s **NOTE:** Since this has two normal 21s and the 21 attached to 14, it is back up to 46 count, the 21 comes after the der(14;21) due to numerical order

4. 46,XX,+13,der(13;14)(q10;q10)
   Female with Robertsonian between 13 and 21and with 2 normal 13s **NOTE:** In this case, the additional 13 comes before the robertsonian translocation because since they are both 13s, it is numerical before structural

5. 46,XY,t(13;15)(q22;q14)
   Male with translocation between 13 and 15, breakpoints at 13q22 and 15q14

6. 46,X,i(X)(q10)
   Female with Turner syndrome variant, derivative chromosome made up of two Xqs (q10) **NOTE:** This abnormal chromosome is an isochromosome

7. 46,XY,inv(1)(p31.1q24)
   Male with inversion of 1, with breakpoints at 1p31.1 and q24: What type of inversion is this?
   NOTE: This inversion is a pericentric (involves both arms)

8. 46,XY,inv(3)(q21.2q24.3)
   Male with inversion of 3, with breakpoints at q21.2 and q24.3: What type of inversion is this?
   NOTE: This inversion is a paracentric (involves one arm)

9. 46,XX,dup(2)(q21.1q32.3)
   Female with tandem duplication of 2, with breakpoints at 2q21.1 and 2q32.3

10. 46,XX,inv dup(2)(q21.1q32.3) or 46,XX,dup(2)(32.3q21.1)
    Female with inverted duplication of 2, with breakpoints at 2q21.1 and 2q32.3 **NOTE:** 2 ways to Write this, one saying inv dup, the other showing the breakpoints inverted

11. 47,XXY
    Male with Klinefelter syndrome

12. 46,XY,t(2;8)(q22;q21.3)
    Male with balanced translocation between 2 and 8, with breakpoints at 2q22 and 8q21.3

13. 46,XY,der(2)t(2;8)(q22;q21.3)
    Female with unbalanced translocation, resulting in derivative chromosome 2 from a translocation between 2 and 8, with breakpoints at 2q22 and 8q21.3

14. 46,XX,der(8)t(2;8)(q22;q21.3)
    Male with unbalanced translocation, resulting in derivative chromosome 8 from a translocation between 2 and 8, with breakpoints at 2q22 and 8q21.3

15. 45,X[14]/46,XX[16]
    Female with mosaic Turner syndrome (14 cells) and a normal cell line (16 cells)

16. 46,X,i(Y)(p10)
    Male with abnormal Y chromosome that only contains two copies of Yp (p10)

17. 46,XY,psu dic(5;7)(q22;p15.3)
    Male with translocation between 5 and 7, where there is only 1 normal 5 and 2 normal 7s, breakpoints are 5q22 and 7p15.3 **NOTE:** there is 2 centromeres on derivative, but the 5 centromere is the active one, therefore it is a psu dic (pseudo dicentric)

18. 48,XXXY
    Male with three Xs and one Y chromosome

19. 46,X,i(Y)(q10)
    Phenotypic female with one X and one derivative comprised of two copies of Yq **NOTE:** This is a phenotypic female because SRY, the start of the sex determination cascade, is in Yp. So even though the patient Y material, it is a female

20. 45,XY,-20
    Male with monosomy 20

21. 46,X,r(X)(p22?1q2?7)
    Female with ring X, questionable breakpoints at Xp22.1 and Xq27

22. 46,XY,add(18)(q22)
    Male with additional material on one 18q with breakpoint within 18q22

23. 46,XY,t(5;20;16)(q22;q13.2;p11.2)
    Male with three way translocation, chromosome 5 breakpoint at q22, chromosome 16 breakpoint at p11.2 and chromosome 20 breakpoint at q13.2. 5qter is moved to 20q, 20qter is moved to 16p, 16pter is moved to 5q

24. 46,XX,add(6)(q21)
    Female with deletion at 6q21 and unknown material after that

25. 47,XY,+18
    Male with trisomy 18

26. 47,XX,+8[8]/46,XX[22]
    Female with mosaic trisomy 8 [8 cells] and normal cell line [22 cells]

27. 46,XX,t(4;12)(p16.1;q24.1)
    Female with translocation between chromosomes 4 and 12, with breakpoints at 4p16.1 and 12q24.1

28. 46,XY,inv(9)(p12q13)
    Male with nothing wrong with his chromosomes except a common population variant inversion 9 (breakpoints at p12 and q13)

29. 46,XY,t(11;22)(q23.3;q11.2)
    A balanced translocation between chromosomes 11 and 22 with breakpoints at 11q23.3 and 22q11.2 in an otherwise normal male

30. 46,X,t(X;11)(p21;p11.2)
    A female with a balanced translocation between X chromosome and chromosome 11 with breakpoints at Xp21 and 11p11.2

31. 48,XXX,+21
    A female with three copies of the X chromosome and three copies of chromosome 21

32. 46,XX,der(9)t(3;9)(q21;q34)
    An unbalanced translocation between chromosomes 3 and 9, with breakpoints at 3q21 and 9q34 in an otherwise normal female. The abnormal chromosome results in a gain of 3qter and a loss of chromosome 9qter

33. 46,XY,der(19)t(X;19)(p22.31;p13.1)
    An male with an unbalanced translocation between X chromosome and chromosome 19, with breakpoints at Xp22.31 and 19p13.1. The abnormal chromosome results in a gain of Xpter and a loss of 19pter

34. 49,XY,+7,8,add(9)(q21),+10
    A bone marrow specimen with trisomy 7, 8, and 10 in a male patient. There is also a deletion of chromosome 9q21 with additional material of unknown origin

35. 47,XX,i(8)(q10),+21
    A female with an abnormal chromosome 8, comprised of two chromosome 8qs. There is also trisomy 21

36. 93,XXYY,+8
    A male POC specimen that has 93 chromosomes with an additional chromosome 8

37. 47,XX,t(11;22)(q23.3;q11.2),+der(22)t(11;22)(q23.3;q11.2)
    Female with a balanced translocation between chromosomes 11 and 22, with breakpoints at 11q23.3 and 22q11.2. There is an additional copy of the abnormal chromosome 22

38. 46,XX,del(8)(p11.2),del(13)(q12q14),inv(16)(p13.1q22)
    A female with a terminal deletion of chromosome 8 at 8p11.2, an interstitial deletion of chromosome 13, breakpoints at 13q12 and 13q14 and an upside down segment of chromosome 16 with breakpoints at 16p13.1 and 16q22

39. 46,X,-Y,+15
    A bone marrow from a male with loss of chromosome Y and trisomy 15 **NOTE:** Since this is an oncology specimen, the loss of Y is noted

40. 46,XY,+1,der(1;7)(q10;p10),der(8;15)(q10;q10),+15,der(21)t(17;21)(p11.2;q11.2)
    A female with a whole arm translocation between chromosomes 1q and 7p that results in trisomy for 1q. This patient also has a whole arm translocation between 8q and 15q that results in trisomy for 15q and an abnormal 21 from a translocation between chromosomes 21 and 17, with breakpoints at 21q11.2 and 17p11.2

41. 46,XY,der(3)t(3;18)(q26.1;q22)
    A male with developmental delay that has an abnormal chromosome comprised from a translocation between chromosomes 3 and 18, with breakpoints at 3q26.1 and 18q22. This abnormal chromosome results in a loss of chromosome 3qter and a gain of 18qter

42. 46,XY,der(13;4)(q10;q10),+15
    A male with a Robertsonian translocation between chromosomes 13q and 14q and trisomy 15

43. 45,X,-X,t(15;17)(q24;q21)
    A female with a translocation between chromosomes 15 and 17, with breakpoints at 15q24 and 17q21 and a loss of X **NOTE:** Since this is an oncology specimen, the loss of X is noted

44. 47,XX,+5,i(5)(p10),del(6)(q21),-18,+mar
    A bone marrow from a female with two normal chromosome 5s and a derivative that is comprised of two chromosome 5p arms, a terminal deletion of 6q at q21, monosomy for chromosome 18 and has an unidentifiable chromosome

45. 46,XX,+13,der(13;13)(q10;q10)
    Normal female sex complement with a normal chromosome 13 and an abnormal chromosome 13 derived from two q arms