**ANSWERS: CHAPTER 28**

**MATCHING**

1. q  8. s  15. r
2. n  9. f  16. d
3. b  10. m  17. o
4. e  11. a  18. j
5. k  12. l  19. h
6. p  13. c
7. g  14. i

**IMAGE LABELING**

1. deletion
2. translocation
3. inversion
4. robertsonian translocation
5. isochromosomal translocation
6. ring formation
7. clubfeet
8. renal anomalies
9. echogenic intracardiac focus (EIF), hypoplastic left heart
10. small, low-set ears
11. brain anomalies, ventricular dilatation
12. facial anomalies
13. polydactyly
14. small omphalocele
15. single umbilical artery
16. shortened limbs
17. spina bifida
18. esophageal atresia
19. small, low-set ears
20. strawberry-shaped head
21. choroid cysts
22. clenched hands, radial aplasia
23. cardiac defects
24. omphalocele
25. single umbilical artery
26. clubfeet
27. widened pelvis
28. pyelectasis
29. cardiac defects, echogenic intracardiac focus (EIF)
30. nuchal thickening
31. midventricular dilatation
32. brachycephaly
33. hypoplastic nose
34. clinodactyly
35. esophageal atresia, duodenal atresia
36. hyperechogenic bowel
37. sandal gap
38. shortened limbs
39. vertebral anomalies
40. tracheoesophageal fistula
41. renal anomalies
42. cardiac defects
43. limb anomalies
44. anal atresia

**MULTIPLE CHOICE**

1. d  6. c  11. d  16. c
2. c  7. d  12. a  17. b
3. a  8. b  13. b  18. c
5. b  10. c  15. a  20. a

**FILL-IN-THE-BLANK**

1a. syndrome
1b. association
1c. sequence
2. DNA
3. germ
4. aneuploidy
5. maternal
6a. infection
6b. drugs
7. maternal serum alphafetoprotein (MSAFP)
8. Craniosyntosis
9a. Edward syndrome
9b. female
10. caudal regression syndrome
11. limb-body wall complex
12. cystic hygroma
13. Pterygium syndrome
14. triploidy
15a. Amniotic
15b. constriction
16a. monosomy X
16b. XO syndrome
17a. coloboma (a hole in the eye structure)
17b. ear anomalies
18. male
19. Aicardi
20a. sperm fertilizing
20b. mother

**SHORT ANSWER**

1. Known risk factors of chromosomal abnormalities are advanced maternal age, previous pregnancy with chromosomal abnormalities, paternal age, history of early pregnancy loss, and ethnicity. Most of these are clearly evident causes of chromosomal anomalies.

2. MSAFP, hCG, unconjugated estriol (uE3), and inhibin-A. The alternative name is quadruple screen.

3. Autosomal dominant: Conditions associated with the 22 nonsex chromosomes are termed autosomal. In autosomal dominant transmission, one parent has the trait or condition, and his or her genetic contribution will “dominate” the genes from the
other parent. When one parent has the dominate gene, there is a 50% chance each child will inherit the trait or condition.

Autosomal recessive: Each parent carries the gene associated with the trait or condition, but is not affected. Each child has a 25% chance of having the condition, a 50% chance of becoming a carrier, and a 25% chance of not inheriting the gene.

Aicardi syndrome: Even though the syndrome is considered to have an autosomal recessive inheritance pattern, the fact that it occurs in females classifies the syndrome as an X-linked dominant inheritance pattern.

Apert syndrome: Because only one copy of the gene is necessary for development of this syndrome, it is considered to be autosomal dominant.

Beckwith-Weidemann syndrome (BWS): Although most cases are sporadic, 10% to 15% are autosomal dominant in nature.

Caudal regression syndrome: This autosomal dominant trait, which has an association with an anterior meningocele, presacral teratoma, and anorectal anomalies, is often referred to as the Currarino triad.

Holt-Oram syndrome (HOS): This is an autosomal dominant mutation resulting from T-box (TBX) gene on chromosome 12.

Noonan syndrome: Noonan syndrome is an autosomal dominant condition affecting males and females equally. This syndrome follows a sporadic occurrence due to mutations on chromosome 12.

4. Apert syndrome: hydronephrosis

Beckwith-Wiedemann syndrome: renal medullary dysplasia, large kidneys, overgrowth of external genitalia, cryptorchidism, Wilms tumor, gonadoblastoma

CHARGE association: central hypogonadism, cryptorchidism, horseshoe kidney, hydronephrosis

Goldenhar syndrome: ectopic and or fused kidneys, renal agenesis, ureteropelvic junction obstruction, multicystic kidney

Triploidy: cystic dysplasia, hydronephrosis, renal hypoplasia, adrenal hypoplasia, hypospadias, cryptorchidism

IMAGEx EVALUATION/PATHOLOGY

1. Marked swelling and edema of the foot are evident (arrows). The curved arrow points to a constriction band of the leg. The pathologic photograph correlates with ultrasound findings, showing marked edema of the food and lower leg secondary to constriction by amniotic band. This is amniotic band syndrome.

2. This surface rendered 3-D construction demonstrates a bulging forehead and upper facial sinking. This fetus was diagnosed with Apert syndrome.

3. This sagittal image of a fetus with LBW complex demonstrates the tethering seen with this group of malformations. Limb-body wall complex is a collection of ventral wall and limb defects. One type of LBWC includes craniofacial defects, and the second type involves short cord, intact amnion, and extra embryonic coelom persistence as well as urogenital malformations, anal atresia, and a meningocele in the lumbosacral region.

4. This image reveals a sagittal view through the head, neck, and torso displaying bilateral hygromas (arrows). Cystic hygromas are congenital lesions housing lymphatic fluid, usually in the neck region. The hygroma can regress leaving a webbed appearance of the neck. Lethal multiple pterygium syndrome, triploidy, trisomy 21, and Turner syndrome may demonstrate cystic hygroma. Many other anomalies such as IUGR, polyhydramnios, short forearms, scoliosis, hypoplastic lungs, and cardiac defects, among others, may be demonstrated with cystic hygroma.

5. The image displays the fetal cranial ventricles, which are dilated bilaterally. Apert syndrome, CHARGE syndrome, and trisomy 21 commonly reflect dilated cranial ventricles.

CASE STUDY

1. This fetal transverse abdomen demonstrates an omphalocele (a type of abdominal wall defect in which the intestines, liver, and sometimes other organs remain outside the abdomen in a sac). The image and symptoms suggest typical pathologic and clinical features of triploidy. BWS, CHARGE syndrome, triploidy, trisomy 13, and trisomy 18 have omphalocele findings.

2. This 11-week fetus demonstrated an abnormally thick nuchal lucency (solid arrow) and an umbilical cord cyst (open arrow). Goldenhar syndrome, monosomy X (Turner or XO syndrome), triploidy, trisomy 13, 18, and 21, lethal multiple pterygium syndrome. A sonographer should interrogate the fetal brain for holoprosencephaly, cystic hygroma, and omphalocele. CVS and/or amniocentesis may offer additional diagnostic information, as well as comfort the parents.