World Database for Pediatric and Congenital Heart Surgery

Appendix E: Chromosomal Abnormalities: Terms and Definitions

11p15.5

11q

12p1.21

12p12.1

12q24

15q21.1

1q42.1

20p12

22q11 deletion
Deletions or mutations involving the long arm of chromosome 22 (critical region 22q11.2) are associated with the DiGeorge sequence, velocardiofacial syndrome, conotruncal face anomaly syndrome, CATCH 22, and some isolated conotruncal malformations.

2p21

3p22

45XOTurner syndrome
(45XO) is a chromosomal deletionabnormality, which occurs in 1:5000 live female births. Although common in first trimester, most 45XO conceptuses are spontaneously aborted. Affected individuals are missing one X chromosome. The major features include short stature, primary amenorrhea due to ovarian dysgenesis, webbed neck, congenital lymphedema, and cubitus valgus. Cardiovascular abnormalities occur in 20-40% of cases, the most common of which is coarctation of the aorta (70%). Additional defects include commissural aortic valve, aortic stenosis, a spectrum of left-sided obstructive defects and/or hypoplastic defects; hypoplastic left heart syndrome, aotic dilation, dissection, and rupture

47XXY
Klinefelter, or 47XXY syndrome, is a sporadic
chromosomal abnormality in which males have at least two X chromosomes and at least one Y chromosome. Incidence is 1:500 males or 1:1000 births. Klinefelter syndrome occurs usually in association with advanced maternal age at conception. It is the most common sex chromosome disorder and the most common cause of hypogonadism and infertility. Cardiovascular abnormalities in more than 50% of cases include mitral valve prolapse, varicose veins and deep venous thrombosis.

4p

4p16

5p

6p12

7q11

7q11.23

7q32

7q34

8q12

TGFBR1 oor 2

Trisomy 08
Trisomy 8, or Warkany syndrome, is a chromosomal abnormality, which is a frequent cause of first trimester spontaneous abortions. Complete Trisomy 8 is usually an early lethal disorder. Incidence is 1:25,000-50,000 births. Affected individuals have an extra (or third) copy of chromosome 8. Cardiovascular abnormalities include septal defects and great vessel anomalies.

Trisomy 09
Trisomy 9 or Rethore syndrome is a rare chromosomal abnormality, which is a frequent cause of first trimester spontaneous abortions. Incidence is 1:100,000 births. Affected individuals have an extra (or third) copy of chromosome 9. Most affected individuals die during infancy or early childhood. Cardiovascular abnormalities occur in 75% of cases and include VSD, ASD, PDA, valve defects, DORV, persistent left SVC, and endocardial fibroelastosis.

Trisomy 13
Patau or Bartholin-Patau syndrome, or Trisomy 13, is a chromosomal abnormality. Incidence is 1:5000-10,000 births. Sporadic cases occur usually in association with advanced maternal age at conception. Affected individuals have an extra (or third) copy of chromosome 13. More than 90% of individuals with Trisomy 13 die within their first days or weeks of life. Only 5-10% survive beyond 1 year of age. Cardiovascular abnormalities in 80% of cases include VSD, PDA, ASD; dextrocardia in more than 50% of cases; and anomalous pulmonary venous connection, overriding aorta, pulmonary stenosis, hypoplastic aorta, mitral valve atresia, aortic valve atresia, and bicuspid aortic valve in fewer than 50% of cases.

Trisomy 18
Edwards syndrome, or Trisomy 18, is a chromosomal abnormality. Incidence is 1:3000-5000 births. Sporadic cases of Edwards syndrome occur usually in association with advanced maternal age at conception. Affected individuals have an extra (or third) copy of chromosome 18. Approximately 50% of infants with Trisomy 18 die within the first week of life, approximately 40% die within the first month of life, only 5-10% survive beyond the first year. Cardiovascular abnormalities in more than 50% of cases include VSD, ASD and PDA; bicuspid aortic and/or pulmonary valves, nodularity of valve leaflets, pulmonic stenosis, coarctation of the aorta in 10-50% of cases; and anomalous coronary artery, TGA, TOF, dextrocardia and aberrant subclavian artery in less than10% of cases.

Trisomy 21

Down syndrome, or Trisomy 21, is the most frequent chromosomal abnormality. Incidence is 1:600-1000 live births. Sporadic cases of Down syndrome occur in strong association with advanced maternal age at conception. Affected individuals have an extra (or third) copy of chromosome 21. Cardiovascular abnormalities in 40-50% of cases, in decreasing order of frequency, include AVSD, VSD, TOF and PDA. Left- sided obstructive defects, such as coarctation and aortic valve stenosis, are rare.