Appendix A, Chromosomal and Structural Anomalies Detected by the Prenatal Screening Program

This brief description of chromosomal and structural anomalies is included for the use of providers and other personnel in the prenatal care office or clinic. Data sources include the California Birth Defects Monitoring Program.

Neural tube defects (NTD)

Neural tube defects (anencephaly, spina bifida and encephalocele) occur in about one in 1,600 births in California. Some studies have shown that Hispanic people are at a slightly higher risk than other ethnic or racial groups. Anencephaly and spina bifida account for approximately 95% of neural tube defects, and encephaloceles make up the remaining 5%. Most neural tube defects are isolated anomalies, but they may also occur in association with a genetic syndrome such as Meckel Gruber syndrome, or with chromosomal abnormalities such as trisomy 18.

Approximately 95% of infants born with neural tube defects are born into families with no previous history of NTDs. This is why prenatal screening is the best method for detecting most open neural tube defects. Any couple with a family history of a neural tube defect should be referred for genetic counseling.

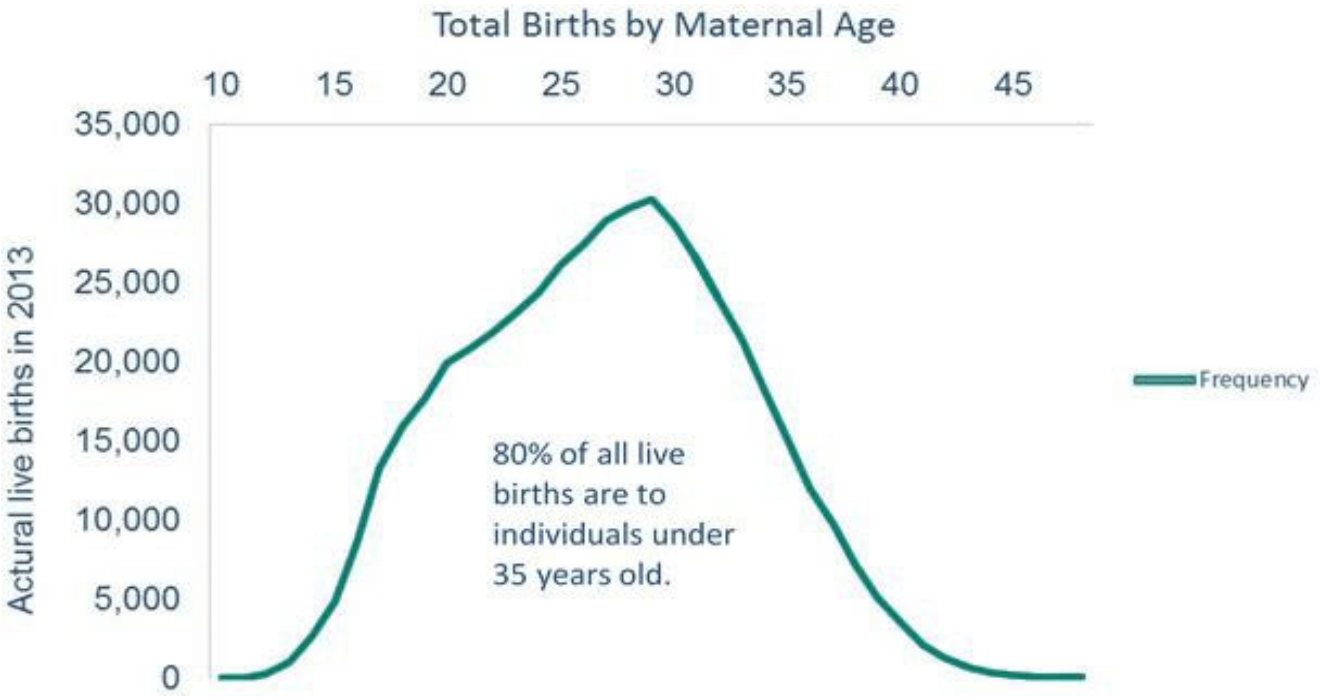
Anencephaly is a defect in which the top of the neural tube fails to close, and the brain does not develop. Anencephaly is always fatal. Seventy-five percent of anencephalic fetuses are stillborn. Twenty-five percent are live born but die within hours or days of birth. Spina bifida (also called meningomyelocele) is a defect in which the neural tube fails to close, and a portion of the spinal cord and nerves fails to develop properly. Spina bifida varies in severity depending upon the size and the position of the defect on the spine, whether it is covered by skin, and the amount of nerve damage.

About 80 to 85% of infants with spina bifida have the more serious form of the condition, an uncovered or open defect. Of these, about 8% are stillborn or die shortly after birth. At least 80% of those who survive to five years of age have a severe handicap (paralysis below the level of the defect, and bowel and bladder incontinence). Many have learning disabilities. 10 to 15% have intellectual disabilities due to associated anomalies of the brain. About 90% develop hydrocephalus, which increases the likelihood of intellectual disabilities if not treated. Prenatal detection of spina bifida, with delivery at a tertiary care medical center, often improves the outcome of these infants.

Note: “Spina bifida occulta” (a defect in a bone of the spine) is not usually considered a neural tube defect. Call your Prenatal Screening Coordinator for more information.

An encephalocele is a sac, usually at the back of the skull, filled with spinal fluid and variable amounts of brain tissue. It carries a high risk for severe neurological deficit and intellectual disabilities. Occasionally other anomalies such as kidney malformations, extra fingers or cleft palate are associated with this neural tube defect. Such findings suggest an inherited genetic disorder (such as Meckel-Gruber syndrome) or trisomy 13 and the couple should be referred for genetic counseling.

Appendix A, continued



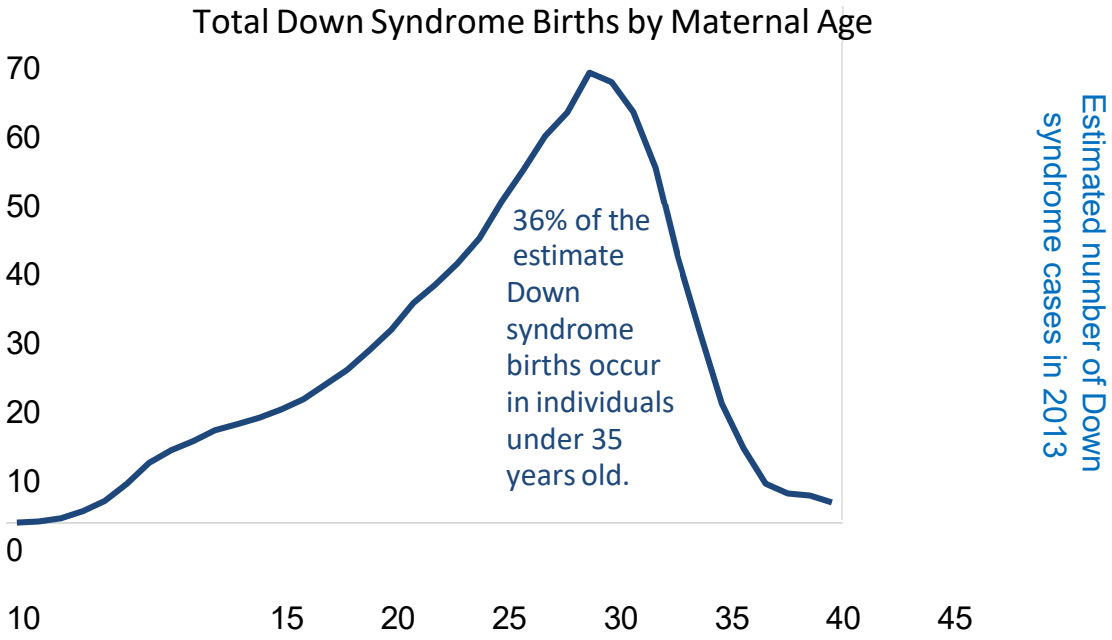
The risk of giving birth to a baby with a chromosomal anomaly, such as trisomy 21, 18, or 13, increases with an individual's age. However, most newborns with these conditions are born to younger individuals because younger individuals, as a group, have more newborns.

Trisomy 21

Down syndrome (trisomy 21), the result of an extra chromosome #21, occurs in approximately 1 in 700 births in California.* The syndrome includes physical characteristics such as short stature, epicanthal folds, flat nasal bridge, and a single crease across the palm. Everyone with this disorder is intellectually disabled, but the degree of disability varies. Most individuals are moderately intellectually disabled.

Approximately 40% also have congenital heart defects. The life span of individuals with Down syndrome varies and is influenced by the presence or absence of congenital heart defects. Anyone with a family history of trisomy 21 (or any other chromosomal anomaly should be referred for genetic counseling.

*Source: California Birth Defects Monitoring Program



Trisomy 18

Trisomy 18 involves an extra chromosome #18. It occurs in approximately 1 in 3,000 births. Approximately 70% of fetuses with trisomy 18 are miscarried before birth. For those who survive to term, 90 to 95% will not reach their first birthday. Those surviving have severe intellectual deficiencies and multiple anomalies including heart and kidney defects. Some have neural tube defects and/or abdominal wall defects. Anyone with a family history of trisomy 18 (or any other chromosomal anomaly) should be referred for genetic counseling.

Trisomy 13

Trisomy 13 involves an extra chromosome #13. It occurs in approximately 1 in every 5,000 births. Approximately 49% of fetuses with trisomy 18 are miscarried before birth. For those who survive to term, 50% die in the first week of life, and 90% die before their first birthday. Those surviving have severe intellectual deficiencies and anomalies including structural problems with the brain, such as the front of the brain not dividing normally (holoprosencephaly) and cleft lip, cleft palate. Anyone with a family history of trisomy 13 or (any chromosomal anomaly), should be referred for genetic counseling.