

INTRO

Edwards syndrome (Trisomy-18) is a chromosomal abnormality characterized by the presence of an extra chromosome 18. Trisomy 18 is the second most prevalent chromosomal disorder to Trisomy-21 (Down syndrome)

The live-born prevalence is estimated as 1/6,000 but the overall prevalence is 1/2500 higher due to the high frequency of fetal loss and pregnancy termination after prenatal diagnosis.



PRENATAL DIAGNOSIS

Mothers can discover possible genetic disorders based on screening by maternal age, maternal serum marker screening, or detection of sonographic abnormalities during the second and third trimesters

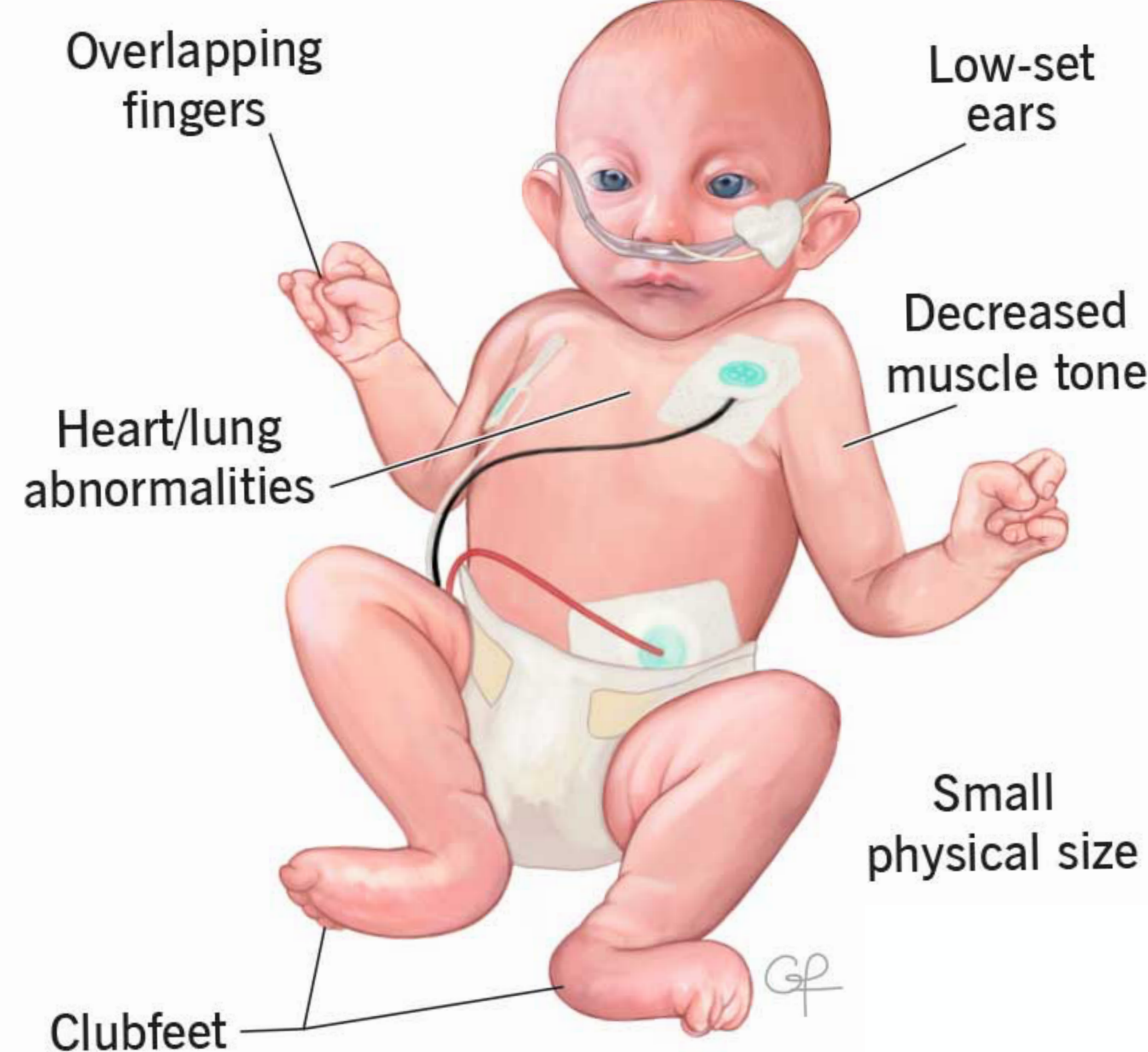
Any woman with results indicating increased chances of having a child with trisomy 18 is offered amniocentesis to ensure fetal well-being.

IDENTIFIERS

Sonogram abnormalities are increased nuchal translucency thickness, growth retardation, choroid plexus cyst, overlapping of fingers, and congenital heart defects.

MAJOR abnormalities found on sonograms are prenatal and postnatal growth deficiency, heightened risk of infant mortality, and cognitive disabilities.

Minor abnormalities seen in sonograms are craniofacial features, clenched fists, small fingernails, short sternum, and underdeveloped thumbs.

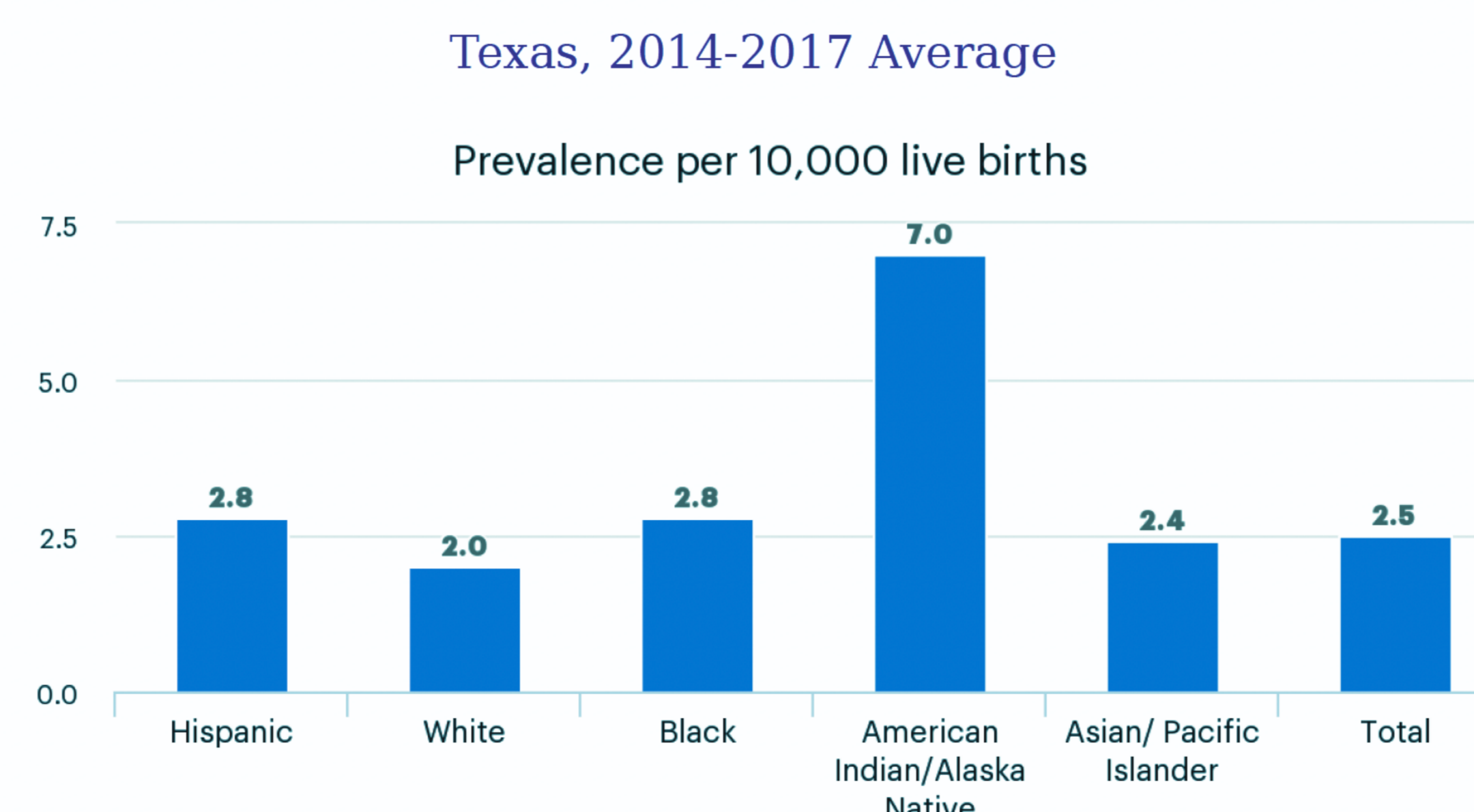


UNRESOLVED QUESTIONS

The controversy surrounding the option of aggressive respiratory, palliative medical guidance, or surgical treatment on infants with trisomy 18.

Due to the high neonatal and infant mortality rate and the quality of life in children with the syndrome, many practitioners in the US and Europe argue for a noninterventionist approach with accompanying comfort care and currently with the guidance of a pacifying care team

TRISOMY 18 BY RACE/ETHNICITY



CHROMOSOMAL DEFECTS

FULL TRISOMY

The fetus has a nondisjunction of two chromosomes 18. All cells are aneuploid leading to increased mortality

PARTIAL TRISOMY

The fetus has only inherited part of an extra copy of chromosome 18. A very rare version of Trisomy 18

MOSAIC TRISOMY

The fetus has inherited a complete extra copy of chromosome 18, but the copy is only present in some cells

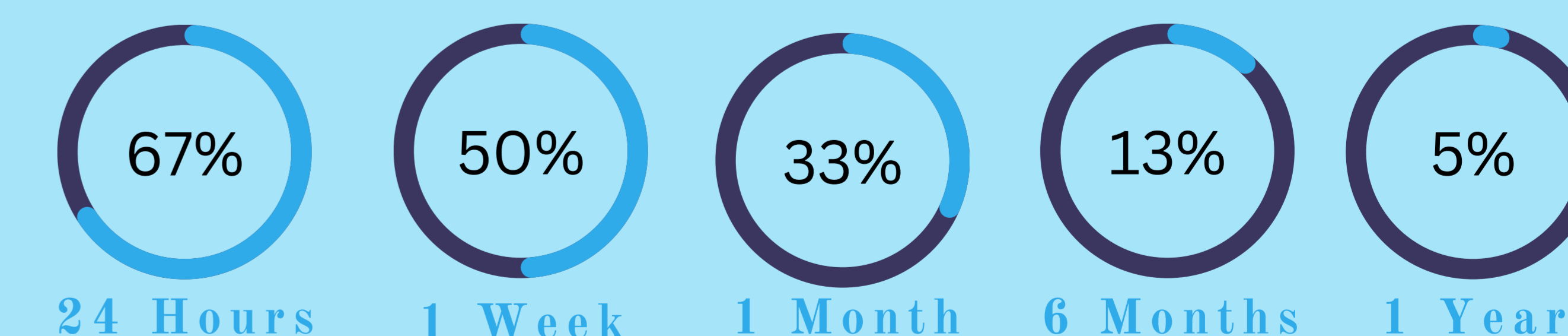
DEMOGRAPHICS

The prevalence of Edwards syndrome is about 1 in 5,000 births. Two-thirds of all newborns with the condition are female because males with trisomy 18 are more likely to be miscarried. Increased parental age is the only known factor shown to heighten the chances of trisomy development. The risk increases with age for both sexes but begins earlier and is more pronounced in women.



MORTALITY

Percent of Infant Mortality



The major causes of death are central apnea, cardiac failure due to cardiac malformations and respiratory insufficiency due to hypoventilation, aspiration, upper airway obstruction, or, likely, the combination of these and other factors