Appendix 12.2

Suggested List of Prenatal Diagnoses That Can Be Included in Prevalence Estimates Without a Clinician's Review of the Defect Certainty

It is recommended that programs engage the services of a clinical geneticist or other consultant knowledgeable about birth defects, fetal development, and prenatal diagnosis to review case information and assess the certainty of all prenatal diagnoses.

If the services of a knowledgeable clinician are not available, it is suggested that inclusion of prenatal diagnoses in prevalence estimates be restricted to the defects on this list; and that prenatal diagnoses not on this list should not be included in defect prevalence estimates.

DEFECT	COMMENT
Abnormal Number of Chromosomes	Karyotype required for diagnosis. Examples include trisomies 13, 18, 21, triploidy, Turner syndrome, Klinefelter syndrome. Note: Some chromosomal abnormalities (e.g., mosaicism) detected by chorionic villus sampling can represent changes in the placenta not present in the fetus and therefore will require confirmation by amniocentesis or postnatally.
Structural Abnormalities:	
Amelia	Absence of the entire limb only. Absence of portions of a limb (e.g., hand only) requires clinical review for inclusion.
Anencephalus	
Cleft lip	
Conjoined twins	
Diaphragmatic hernia	
Encephalocele	
Heart defects:	
Atrioventricular septal defect	
Ebstein's anomaly	
Hypoplastic left heart syndrome	
Single ventricle	
Sacral agenesis	
Sirenomelia	
Spina bifida	