UNC SCHOOL OF MEDICINE

UNC Center for Maternal & Infant Health

Prenatal Population Screening
For Common Autosomal Recessive Disorders

Ethnicity	Common Conditions	Carrier Frequency	Testing Methods	Detection Rate	Recommended by ACOG*	Recommended by ACMG**
Caucasian	Cystic Fibrosis	1/25	Molecular	88%	X	X
Ashkenazi Jewish	Cystic Fibrosis	1/24	Molecular	94%	X	X
	Tay-Sachs	1/31	Molecular & Enzyme(Hex-A)	92-99% 98%	X	X
	Canavan	1/41	Molecular	97.4%	X	X
	Familial Dysautonomia	1/31	Molecular	>99%	X	X
	Fanconia anemia group C	1/89	Molecular	>99%		X
	Niemann-Pick disease type A	1/90	Molecular	97%		X
	Mucolipidosis IV	1/127	Molecular	95%		X
	Bloom Syndrome	1/107	Molecular	>99%		X
	Gaucher's disease	1/18	Molecular	89-95%		X
French Canadian/ Creole	Tay-Sachs	1/30	Enzyme (Hex-A)	98%	X	X
African American	Sickle Cell Trait Other Hemoglobinopathies	1/12 >1/30	Hemoglobin electrophoresis See flow chart	Variable	X	X
Mediterranean, Southeast Asian* **	Hemoglobinopathies	>1/20	See flow chart	Variable	X	X

*ACOG: American College of Obstetrics and Gynecology
**ACMG: American College of Medical Genetics
*** Southeast Asian ancestry: Burma, Brunei, Cambodia, China, East Timor, Indonesia, Laos, Macau, Malaysia, Mongolia, Phillipines, Singapore, TaiwanThailand, Vietnam
Mediterranean ancestry: Cyprus, Greece, Italy, Portugal, southern Spain

Prenatal Evaluation for Hemoglobinopathies: (click here for algorithm)

References

1. ACOG Committee Opinion, Number 298, August 2004: Prenatal and Preconceptional Carrier Screening for Genetic Diseases in Individuals of Eastern European Jewish Descent.
2. ACOG Committee Opinion, Number 325, December 2005: Update on Carrier Screening for Cystic Fibrosis.
3. ACOG Practice Bulletin, Number 78, January 2007: Hemoglobinopathies in Pregnancy.
4. ACMG Practice Guidelines, January 2008: Carrier Screening in individuals of Ashkenazi Jewish Descent.
5. NSGC Publication, 2005: Ancestry Based Carrier Screening.

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Notification to Users

These algorithms are designed to assist the primary care provider in the clinical management of a variety of problems that occur during pregnancy. They should not be interpreted as a standard of care, but instead represent guidelines for management. Variation in practices should take into account such factors as characteristics of the individual patient, health resources, and regional experience with diagnostic and therapeutic modalities.