

Birth Defects Risk Factors–Maternal Conditions

Maternal Condition	Potential Fetal Complications	Incidence	Maternal Prevention Strategies
Diabetes Mellitus	Multiple congenital malformations including: caudal regression syndrome, NTD, cardiac defects, microcephaly, hydronephrosis, renal and ureteral defects, duodenal or anorectal atresia	Major congenital malformations found in 5-9 % of infants born to mothers with poorly managed existing diabetes ^{1, 2}	T Maintain normal blood sugar levels before conception and throughout pregnancy
Seizure Disorders	NTD, facial defects, hypoplasia of distal phalanges, hypertelorism	Approximately 6-8 % risk of birth defects among infants born to mothers receiving anticonvulsant therapy; no consensus as to what medication is most teratogenic to a developing fetus ^{3, 4, 5, 6}	T Evaluate potential teratogenicity of seizure medications before conception T Adjust medications or dosage before conception to maintain seizure control (lowest dose of a single drug is preferable)
Systemic Lupus Erythematosus (SLE)	Congenital heart block, fetal death, serologic abnormalities, skin lesions	3 % of infants born to mothers with SLE present with complications ⁷	T Close monitoring of lupus-related laboratory values T Serial fetal ultrasound studies to assess growth T Assessment of fetal functioning T Adjustment of corticosteroid medications to optimize maternal health

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Metabolic Disorders: Phenylketonuria	Mental retardation, microcephaly, congenital heart disease	<p>Among women with PKU on unrestricted diets:</p> <p>risk of mental retardation = 92 %</p> <p>risk of microcephaly = 73 %</p> <p>risk of cardiac defects = 12 %</p> <p>risk of PKU in offspring of woman with PKU: 1/120 ^{8, 9, 10}</p>	<p>T Specialized medical/nutritional management of woman prior to conception and throughout pregnancy</p> <p>T Maintain contact with females diagnosed with PKU through newborn screening to keep patients informed of implications for childbearing and available strategies to reduce risk</p>
Alcohol Abuse	Growth deficiencies, mental retardation, developmental delays, fine-motor dysfunction, poor attention span, speech problems, behavioral/psychosocial/cognitive problems, craniofacial and skeletal anomalies, cardiac defects	Fetal Alcohol Syndrome (FAS): 1/750 to 1/1000 live births ^{11, 12}	<p>T Avoid alcohol prior to conception and throughout pregnancy</p>

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