DOWN SYNDROME –OSCE TEST YOURSELF

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26 yr old women is just dx as pregnant . she is concerned about her pregnancy because one of her relative has a child with down syndrome. She few querries regarding early dx of down syndrome

- **Which is the most sensitive test for down syndrome in first trimester?**
- **Name four soft sonologic signs in 2nd trimester.**
- **Compare the detection rate of ist trimester and 2nd trimester screening tests.**
- **Name 4 foetal anomalies associated with aneuploidy.**

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Selected Down Syndrome Screening Strategies and Their Detection Rate

Strategy	Analytes	Detection Rate[a] (%)
First-trimester screen	NT, PAPP-A, and hCG or free β-hCG	79–87
NT	NT alone	64–70
Triple test	MSAFP, hCG or free β-hCG, uE3	61–70
Quadruple (Quad) test	MSAFP, hCG or free β-hCG, uE3, inh	74–81
Integrated screen	First-trimester screen and Quad test; results withheld until Quad test completed	94–96
Stepwise sequential screen	First-trimester screen and Quad test 1% offered diagnostic test after first-trimester screen 99% proceed to Quad test, results withheld until Quad test completed	90–95
Contingent sequential screen	First-trimester screen and Quad test 1% offered diagnostic test after first-trimester screen 15% proceed to Quad test; results withheld until Quad test completed 84% have no additional test after first-trimester screen	88–94
Cell-free fetal DNA testing (high-risk pregnancies)	No analytes—massively parallel genomic sequencing	98

Second-Trimester Sonographic Markers or "Soft Signs" Associated with Down Syndrome Fetuses

Sonographic Marker[a]
Brachycephaly or shortened frontal lobe
Clinodactyly (hypoplasia of the 5th digit middle phalanx)
Echogenic bowel
Flat facies
Echogenic intracardiac focus
Nasal bone absence or hypoplasia
Nuchal fold thickening
Renal pelvis dilation (mild)
"Sandal gap" between first and second toes
Shortened ear length
Single transverse palmar crease
Single umbilical artery
Short femur
Short humerus
Widened iliac angle

[a]Listed alphabetically.

Aneuploidy Risk Associated with Selected Major Fetal Anomalies

Abnormality	Birth Prevalence	Aneuploidy Risk (%)	Common Aneuploidies[a]
Cystic hygroma	1/5000	50–70	45,X; 21; 18; 13; triploidy
Nonimmune hydrops	1/1500–4000	10–20	21, 18, 13, 45X, triploidy
Ventriculomegaly	1/1000–2000	5–25	13, 18, 21, triploidy
Holoprosencephaly	1/10,000–15,000	30–40	13, 18, 22, triploidy
Dandy-Walker malformation	1/12,000	40	18, 13, 21, triploidy
Cleft lip/palate	1/1000	5–15	18, 13
Cardiac defects	5–8/1000	10–30	21; 18; 13; 45,X; 22q11.2 microdeletion
Diaphragmatic hernia	1/3000–4000	5–15	18, 13, 21
Esophageal atresia	1/4000	10	18, 21
Duodenal atresia	1/10,000	30	21
Gastroschisis	1/2000–4000	No increase	
Omphalocele	1/4000	30–50	18, 13, 21, triploidy
Clubfoot	1/1000	5–30	18, 13