

QUICK REFERENCE GUIDE - OPTIMAL PRENATAL GENETICS SPECIMEN REQUIREMENTS

TEST CATEGORIES	AMNIOTIC FLUID	CHORIONIC VILLI SAMPLING (CVS)
Chromosomal Microarray Analysis (CMA) AND Chromosome Analysis (Limited or Full) + AFP +/- Aneuploidy FISH*	30 cc in two or three 15 cc screw-top polypropylene tubes (Discard first 2 cc) Cultured cells not accepted OPTIMAL GESTATIONAL AGE > 17 wks	30-35 mg in 15 cc screw-top tube with sterile transport media Cultured cells not accepted
Chromosomes / AFAFP AND Aneuploidy FISH	20-25 cc in two 15 cc screw-top polypropylene tube (Discard first 2 cc)	25-30 mg in 15 cc screw-top tube with sterile transport media
Chromosome Analysis AND AFAFP ONLY	15-20 cc in two 15 cc screw-top polypropylene tubes (Discard first 2cc)	15-20 mg in 15 cc screw-top tube with sterile transport media
Chromosomal Microarray Analysis (CMA) ONLY*	15-20 cc in two 15 cc screw-top polypropylene tubes (Discard first 2cc) Preferred sample is direct AF, however cultured amniocytes accepted: 1 T25 flask at ~80% confluence. OPTIMAL GESTATIONAL AGE > 17 wks	15-20 mg in 15 cc screw-top tube with sterile transport media Preferred sample is direct CVS, however cultured CV accepted: 1 T25 flask at ~80% confluence.
Chromosomal Microarray Analysis (CMA) AND/OR Chromosome Analysis + AFAFP AND Molecular/Biochemical testing	30 cc in two or three 15 cc screw-top polypropylene tubes (Discard first 2 cc) Call to discuss case before sending. OPTIMAL GESTATIONAL AGE > 17 wks	30-35 mg in 15 cc screw-top tube with sterile transport media Call to discuss case before sending.
Single Gene Molecular/Biochemical testing ONLY	Call to discuss before sending.	Call to discuss before sending.
Prenatal Noonan Panel* Prenatal COL1A1 & COL1A2 Panel*	If other prenatal studies are to be performed at the MGL, send at least 30 ml of AF total for all testing. If another lab is culturing the fetal specimen, send 1 T25 flask at approximately 80% confluence, and 1 T25 flask at ~80% confluence to follow if confirmation studies are necessary.	If other prenatal studies are to be performed at the MGL, send at least 30 mg of CVS total for all testing. If another lab is culturing the fetal specimen, send 1 T25 flask at approximately 80% confluence, and 1 T25 flask at ~80% confluence to follow if confirmation studies are necessary.

*5cc EDTA blood samples from both parents are required.

SPECIAL CONSIDERATIONS FOR PRENATAL CMA SAMPLES:

Amniotic Fluid Samples collected before 16 weeks gestation cannot be used for Direct DNA prep - such samples will be cultured
Cultured fetal specimens >3 weeks old may yield uninterpretable results

SUBOPTIMAL SPECIMENS LIKELY TO CAUSE LONGER TURN AROUND TIMES:

AF sample collected < 16 wks or > 26 wks	AF pellet with RBC present or micropellet
AF sample < 15 ml volume	Cystic Hygroma Fluid
Bloody or discolored AF sample	CVS < 10 mgs, after cleaning