

RECOMMENDED APPROACH FOR PATIENTS WITH ELEVATED MSAFP

Approximately 1% of patients having screening for Down syndrome by SIPS/IPS/Quad are found to have an elevated maternal serum AFP (MSAFP) resulting in a positive screen for open neural tube defect. A review of close to 600 of cases of MSAFP with AFP >2.5MoM (positive screen for ONTD) seen in Medical Genetics over a period of 6 years shows that only 9% of those cases had a fetal structural abnormality related to the elevated MSAFP (spina bifida, anencephaly, omphalocele, gastroschisis, limb body wall complex). Another 5% had IUGR with or without oligohydramnios or echogenic bowel likely indicative of abnormal placentation. 5% of patients had fetal abnormalities unrelated to the elevated MSAFP, 3% had isolated echogenic bowel, 2% had other soft markers and 2% had fetal demise. As such, 73% had completely normal ultrasounds.

Based on these findings, we no longer recommend that all patients with elevated MSAFP be referred to Medical Genetics. Instead, only patients with extremely high MSAFP (≥ 400 $\mu\text{g/L}$) should be referred to Medical Genetics prior to doing any additional investigations. All other patients should have an ultrasound done in their local community as soon as possible even if less than 19 weeks gestation. The result of that ultrasound should be used to guide further management as follows:

1. Patients with fetal structural abnormality should be referred to the Fetal Diagnosis Service at BCWH: <http://www.bcwomens.ca/our-services/pregnancy-prenatal-care/complications-in-pregnancy/fetal-diagnosis-service>; or to the Antenatal Assessment Unit in Victoria (fax referral to 1-250-727-4441)
2. Patients with fetus with AC or EFW less than the 10th %ile should be referred to an MFM specialist as per MFM Provincial Guideline: <http://www.bcwomens.ca/health-professionals/refer-a-patient/ultrasound>
3. Patients with soft markers on ultrasound should be managed as per the recommendations outlined in the PSBC Obstetric Guideline: Prenatal screening for Down syndrome, appendix 4 available at www.bcprenatalscreening.ca
4. Patients with no fetal abnormality identified on ultrasound but with incomplete anatomical screen (not all details seen) should have a repeat ultrasound between 19-21 weeks gestation.
5. All patients with MSAFP that is not explained by a fetal abnormality should be considered at increased obstetrical risk and followed as per following algorithm:

