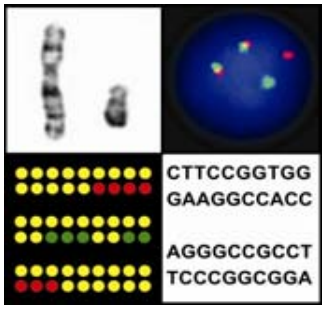

UW Cytogenetic Services

Wisconsin State Laboratory of Hygiene

Spring 2015

Introducing: Smart Start Prenatal Screening

Non-invasive prenatal screening is increasingly becoming more common in today's OBGYN clinics. This testing analyzes fetal cell-free DNA fragments obtained from maternal blood specimens to screen for fetal aneuploidy (Down syndrome, Trisomy 18, etc.) and discrete microdeletion syndromes. Currently, there are several companies offering this clinical screening test, one of which is Illumina (Verifi Prenatal Test).

The UW Cytogenetics Laboratory has recently become a lab partner with Illumina. This partnership allows clinicians to order this testing directly through our laboratory. The UW Cytogenetics Laboratory is calling this testing option the **Smart Start Prenatal Screen** and it is available immediately.

The Smart Start Prenatal Screening test screens for common fetal aneuploidies (trisomy 21, 18, 13), and will include additional options for screening for sex chromosome aneuploidies, trisomies 9 and 16, and certain microdeletion syndomes. The microdeletion panel includes chromosome 22q11 deletion (DiGeorge/Velocardiofacial syndrome), 15q11 deletion (Angelman/Prader-Willi), 1p36 deletion, 4p- (Wolf-Hirschhorn), and 5p- (Cri-du-chat).

Patient consent and clinician signature will be *required* for this testing. As with our other test offerings, the UW Cytogenetics Laboratory can either utilize institutional billing or bill directly to the patient's insurance. The Smart Start Prenatal Screen can be ordered through our new prenatal test requisition form (see below).

Please call our laboratories at 608-262-0402 with any questions.

New Prenatal Test Requisition Form

Over the past couple years the UW Cytogenetic Laboratory has been increasing its prenatal test offerings, first with prenatal microarray testing and now with the Smart Start Prenatal Screening test (see above). Given this increase in test offerings and the specialized nature of the information requested to do this testing, the UW Cytogenetic Laboratory is implementing an additional test requisition form that will be for prenatal testing only. This new requisition form will include the same routine information requested on our current genetic diagnosis requisition form, but with additional areas designated for patient consent signatures and clinician signatures, both of which will be required for the Smart Start Prenatal Screening. The new prenatal test requisition form can be found on our website http://www.slh.wisc.edu/clinical/cytogenetics/ or by calling our laboratory at 608-262-0402.

UW Cytogenetic Services

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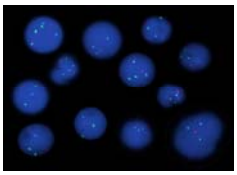

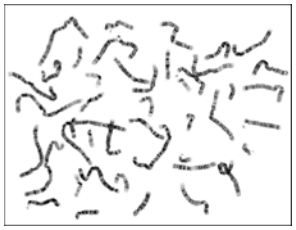

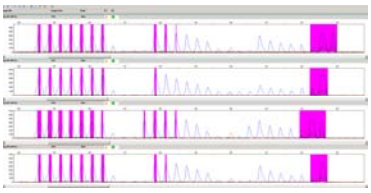

Disease Spotlight: 22q11.2 Microdeletion Syndrome

22q11.2 microdeletion syndrome (which includes DiGeorge syndrome, velocardiofacial syndrome, and Shprintzen syndrome) is caused by a small (submicroscopic) deletion occurring in the long arm of chromosome 22. The features seen in each individual with a 22q11.2 microdeletion syndrome can vary greatly, even within the same family.

Features of 22q11.2 microdeletion syndrome

Some of the most common features of 22q11.2 microdeletion syndrome include:

- Congenital heart disease
- Palatal abnormalities, including cleft palate and velopharyngeal incompetence
- Learning difficulties
- Immune deficiency

Additional features can include hypocalcemia, renal anomalies, gastrointestinal problems, hearing loss, seizures, skeletal anomalies, autoimmune disorders, autism, and psychiatric illness. Developmental delay and intellectual disability can also been seen in children with 22q11.2 microdeletion syndrome.

Testing for 22q11.2 microdeletion syndrome

Testing for 22q11.2 microdeletion syndrome can be done either by fluorescence in situ hybridization (FISH) testing or chromosomal microarray analysis. Rarely, a person with 22q11.2 microdeletion syndrome will have a normal FISH result due to atypical deletions; chromosomal microarray should detect both typical and atypical deletions.

Testing for 22q11.2 microdeletion syndrome should be considered in the following instances:

- Anyone who has clinical features consistent with this microdeletion syndrome.
- Any person with a congenital heart defect, especially conotruncal defects such a tetralogy of Fallot.
- Any newborn suspected of having a 22q11.2 microdeletion by non-invasive prenatal screening (ex. Smart Start Prenatal Screening).

Although approximately 95% of 22q11.2 deletions are *de novo,* the features of 22q11.2 microdeletion syndrome can be relatively mild and differ within family members. Therefore, Parental testing can also be performed if a child is found to have a 22q11.2 deletion.

Please call our laboratory at 608-262-0402 with any questions regarding 22q11.2 microdeletion syndrome or its associated testing.