



MICRODELETION

panel



*...because you want
to know **more**
about the health
of your baby*

www.genetics.emory.edu



Emory Genetics Laboratory offers a Microdeletion Panel that tests for **eight additional conditions** that may not be detected on routine chromosome analysis. The Microdeletion Panel can be performed in conjunction with most prenatal testing procedures, such as chorionic villus sampling and amniocentesis.



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benefits of the microdeletion panel



The **Microdeletion Panel** allows detection of 8 additional conditions that cause learning disabilities/mental retardation and birth defects that are not always detected on routine prenatal testing. A standard chromosome analysis will detect missing chromosomes (such as Turner syndrome) or extra chromosomes (such as Down syndrome). However, very small missing (called a microdeletion) or extra (called a microduplication) chromosome material is usually not detected by standard chromosome analysis. Several microdeletion syndromes have been identified on a variety of chromosomes. Microdeletions can be detected by using a special laboratory test called fluorescence *in-situ* hybridization or FISH.

Conditions Included in the Microdeletion Panel

Individuals with **Angelman syndrome** may have severe mental retardation, seizures, low muscle tone, an unusual walking pattern, inappropriate laughter, as well as other symptoms.

Cri du chat syndrome is so named because infants with this condition have a cry that sounds similar to a cat. Children may also have a small head, a small chin, low muscle tone and severe mental retardation.

Miller-Dieker syndrome is characterized by slow growth even before birth, heart defects, cataracts and a small head. On special imaging tests, these children have a smooth brain. Because of the brain abnormalities, these children have severe mental retardation.

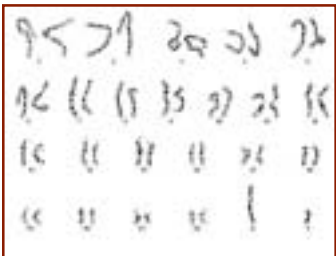
Individuals with **Prader-Willi syndrome** are usually born with an average weight and length. Because of muscle weakness, these children often have difficulty eating at birth and for several months after birth. When muscle tone improves, the children are unable to control their eating because the brain does not get the proper message that the child is full and should stop eating. Therefore, these children may rapidly develop morbid obesity. Most have mental retardation and may also have seizures, sleep disturbances, behavioral problems as well as other characteristics.

Smith-Magenis syndrome is associated with congenital heart defects, mental retardation, self-destructive behavior (such as head banging) as well as other characteristics.

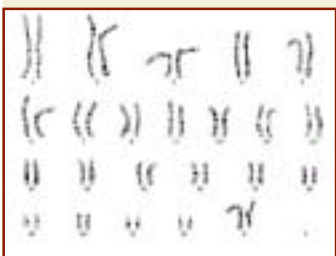
Velocardiofacial (VCFS) and DiGeorge (DG) syndromes have overlapping symptoms. Many individuals have congenital heart disease (74%) and immune deficiency (77%). Other symptoms include significant feeding problems (30%), kidney anomalies (37%), hearing loss, cleft palate, growth hormone deficiency, seizures, schizophrenia, dental problems and other health problems. In some instances this condition is inherited from a parent who may have very mild or few symptoms. If an unborn child is found to have this syndrome, the severity of the condition cannot be determined prior to birth.

Individuals with **Williams syndrome** are usually small before birth. These children usually have very outgoing personalities and may be quite talkative, but most have mental retardation. Heart defects, attention deficit disorder, and hypersensitivity to sound are among other characteristics present in many children with this disorder.

Wolf-Hirschhorn syndrome is characterized by congenital heart defects, seizures, mental retardation, poor growth as well as other features.



46,XY, normal male karyotype
(chromosome analysis).



46,XX, normal female karyotype
(chromosome analysis).

The **Microdeletion Panel** uses FISH to detect the presence or absence of a microdeletion on a given chromosome. However, for some conditions, such as Angelman and Prader-Willi syndromes, there are other causes of the condition that are not detected by FISH testing. Therefore, the **Microdeletion Panel** only detects cases of Angelman syndrome and Prader-Willi syndrome caused by an actual deletion of chromosome material. The following table lists the chromosome involved, how common each condition is (incidence) and how often the **Microdeletion Panel** will detect the condition (detection).

limitations of the microdeletion panel

Chromosome	Microdeletion Syndrome	Incidence	Detection (%)
15	Angelman Syndrome	1 in 20,000 to 1 in 50,000	70
5	Cri du chat syndrome	1 in 20,000 to 1 in 50,000	>99
17	Miller-Dieker	1 in 100,000	>99
15	Prader- Willi syndrome	1 in 10,000	70
17	Smith-Magenis syndrome	1 in 25,000	>99
22	VCFS and DG syndromes	1 in 4,000	>90
7	Williams syndrome	1 in 10,000	>99
4	Wolf-Hirschhorn syndrome	1 in 95,000	>99

The **Microdeletion Panel** is intended to be used to screen for additional conditions which cause learning disability/mental retardation and birth defects. However, if there *continued on top of next page*



Please read this information carefully and feel free to ask any questions prior to signing the form.

I, _____, consent/agree to the following:

1. This testing is performed in addition to routine chromosome analysis. The risks and benefits of the prenatal procedure (CVS, amniocentesis, etc.) have been explained to me previously by _____. The Microdeletion Panel must be ordered in addition to routine chromosome analysis.
2. The Microdeletion Panel does not detect every case of these microdeletion syndromes. Therefore, it is possible to have a child with one of these syndromes, even though the Microdeletion Panel results are normal. In addition, I could have a child with mental retardation and/or birth defects from other causes.
3. By signing this form, I acknowledge that I have had the opportunity to ask questions.
4. The results of the Microdeletion Panel may be unclear or require testing of other family members for accurate interpretation.
5. Because of the complexity of this testing, results will only be given to me by a physician or genetic counselor.
6. Participation in this testing is voluntary.

continued. . .

CONSENT FORM

is a family history of one of these conditions, we recommend genetic counseling to determine the best testing method. Also, ultrasound findings may indicate a more limited FISH panel or other targeted genetic testing. This panel is **not** a substitute for routine chromosome analysis.

Other Considerations

Although rare, this panel may detect microduplications in the chromosome regions studied. Microduplications may lead to mental retardation and other health problems. In the event a microduplication is detected, genetic counseling is available to discuss the results.

This testing is optional, and is not covered by most insurance companies as routine or standard of care. Therefore, if you elect this testing, you must understand that you are responsible for charges at the time of sample collection. The cost of the testing is \$650.

Because of the complexity of this testing, we **strongly recommend** genetic counseling prior to proceeding with testing. Genetic counselors at Emory University are available for consultation by calling (404) 297-1500. **A signed consent form is required before testing will be performed.**



7. I can withdraw my consent for testing at any time. If I withdraw my consent within 24 hours of ordering the test, there will be no cost to me. After that time, I will be charged.

8. I understand that I am responsible for payment for the Microdeletion Panel at the time of specimen collection, and that Emory Genetics Laboratory will not submit a claim for this testing to my insurance company. I understand my insurance company may not cover the cost of this testing, should I decide to file for reimbursement on my own.

Signature: _____ Date: _____

Billing Information:

Make checks payable to Emory Genetics Laboratory

Emory Genetics Laboratory accepts Visa and Mastercard.

Credit Card Number _____

Expiration Date: _____

I authorize Emory Genetics Laboratory to charge \$_____ to my (check one) Visa Mastercard

Authorized Signature _____

If you require a receipt, please contact our billing department at (404) 297-1500.