Elements of Consent for Genetic Testing: Chromosomal Microarray Analysis (CMA)

The potential implications of genetic testing differ from other clinical tests and, therefore, require informed consent. The elements of the consent process are similar to other studies or procedures (describing the test, limitations, risks, and benefits to the patient). However, there are also specific disclosures that are important for the patient/parent to understand. The basic elements below are taken from the Texas Medical Association. (https://www.texmed.org/Template.aspx?id=1745)

Discussion

Informed consent begins with a discussion of the test. The following issues should be discussed when obtaining consent for Chromosomal Microarray Analysis (CMA) for evaluation of developmental delay. The provider may also choose to use educational materials that help to explain CMA and basic genetic principles to a patient/parent.

- Differential diagnosis
  - The child has exhibited signs of developmental delay, and the exact reason is unknown. One possible cause is a genetic abnormality (the provider may wish to refer to **Non-Genetic Causes of Developmental Delay** document).
- Description and purpose of the test
  - Chromosomal Microarray Analysis is a comprehensive technique by which chromosomes are analyzed for copy number variants, in other words, small deletions or duplications of genetic material. Abnormalities detected by CMA can reveal an underlying genetic cause for the child's developmental delay (the provider may wish to refer to **CMA** document).
- Benefits and expected outcome
  - Determining a diagnosis helps the family and their providers better understand the child's condition and can help to guide management and treatment decisions. Sometimes, results of a CMA help to identify the cause of the diagnosis and to predict whether it may affect other individuals in the family (the provider may wish to refer to the **CMA** document).
  - CMA results can be positive, meaning that one or more small deletions or duplications of genetic material were found that may explain the patient's developmental delay or other clinical symptoms. Alternatively, results can be negative, meaning that no deletions or duplications were found. However, it is still possible that the patient's delay may have a different genetic cause. There can also be chromosomal variations of uncertain significance. This means that a change in genetic material was identified but it is unclear whether it explains or causes any clinical symptoms.
- Risks associated with testing (see **CMA** document)
  - Sometimes, a CMA may show that a child has very similar copy number variants on the chromosomes they received from their father as they do on

the chromosomes they received from their mother. This result indicates that perhaps the child's parents are related by blood, for instance as cousins.
    - If a deletion or duplication is found in the child, testing of the parents is typically recommended as well. If one of the parents has the same deletion or duplication and yet does not have developmental delay or the same clinical symptoms as the child, it is less likely that the deletion or duplication caused the child's symptoms. By contrast, if the deletion or duplication occurred anew in the child, it is more likely to explain his/her clinical symptoms. It is important to note that, in doing parental testing, it is sometimes possible to identify misattributed paternity. In other words, the father who was tested is not the biological father of the child.
- Alternatives to testing
    - The patient/parent can choose not to have a CMA done. In this case, the child may still benefit from referrals for developmental therapies to try to address the symptoms of developmental delay.
    - To further evaluate for the underlying cause of the delay and to better understand the possible chance for other family members to be affected, the family may still benefit from referrals to Genetic or Developmental Pediatric specialists, even if they do not wish to pursue genetic testing.
    - Other biochemical tests, genetic tests and imaging studies may also be considered.
- Consequences of no testing
    - Without having a CMA or other tests done, the cause of the child's developmental delay may go unknown. In some cases, there may be a treatable cause that is not identified without testing. There may also be additional health problems for which the child may be at risk that are not identified without testing. Additionally, it may not be possible to determine whether other family members may be at risk for related symptoms.
    - Treatments and therapies aimed at the child's symptoms may be beneficial but also may not address the child's specific problem.

Education

- See available hand outs

Obtaining Written Consent

- Each lab has its own consent form for CMA testing. Your institution may also have a consent form that you need to complete.

Chart Documentation

- The informed consent process should be documented in the medical record.

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This document has been prepared for the use of healthcare professionals when advising patients and families about genetic testing. This does not constitute a legal opinion. If a legal opinion is required, referral to a qualified attorney is recommended.