MOLECULAR DIAGNOSIS for X-LINKED INTELLECTUAL DISABILITY
WHAT IS X-LINKED INTELLECTUAL DISABILITY?

Intellectual disability (ID), sometimes called mental retardation (MR) or developmental delay (DD), is diagnosed when a child has below average scores on intelligence tests and has trouble performing everyday tasks. They may learn and develop more slowly than other children. The degree of intellectual disability can vary significantly, from mild to profound.

There are many causes of intellectual disability, and most of the time the exact cause for ID in a specific child is never known. Some causes of ID are environmental and can include infections in the mother while pregnant, infections in the child as an infant, a serious head injury, or malnutrition. In other cases, the cause may be genetic, which means it is caused by changes in the child’s genes (DNA). X-linked intellectual disability (XLID) is one type of genetically caused ID.

The term “X-linked” means that the gene for the condition is on the X chromosome. Males have one X chromosome and females have two X chromosomes. If a male has a mistake in one of the genes on the X chromosome, he will have symptoms of the condition. If a female has a mistake in one of the genes on one of her X chromosomes, she will either have no symptoms or have mild symptoms of the condition, since the gene on the other X chromosome does not carry a mistake. Females can, however, pass the change on to their sons, who would then have symptoms of the condition. These females who carry a copy of the gene with the mistake are called “carriers”.

SYNDROMIC VERSUS NONSYNDROMIC

If a boy has medical problems in addition to intellectual disability, his condition is said to be “syndromic”. Medical problems could include heart defects, hearing loss, problems with growth, behavior problems, or vision problems.
If a boy has intellectual disability without additional medical problems, the ID is said to be “nonsyndromic”. Syndromic and nonsyndromic intellectual disability are usually caused by mistakes in different genes.

**GENETIC TESTING FOR XLID**

Genetic testing is available for some but not all forms of XLID. Genetic testing is useful in trying to identify the cause of the XLID present in a family and to predict what other health problems, if any, an affected child might develop. Genetic testing for XLID is a blood test and generally takes about 16 weeks for results. If the cause of XLID can be determined, then women in the family can be tested to see if they are at risk for having sons with XLID. Prenatal genetic testing would also be available.

If genetic testing for XLID is negative, meaning that a genetic cause was not found, it does not necessarily mean that the child does not have a form of XLID. It just means that he does not have a form of XLID caused by a gene that has been identified. New genes involved in XLID continue to be discovered. Additional genetic testing in the future may be able to identify a genetic cause for the XLID in a child who tests negative now.

Sometimes it is difficult to interpret genetic test results. In these cases, testing of the child’s mother and other family members may be recommended in order to try to understand the child’s results.
**WHAT NEXT?**
If you are concerned your child may have XLID, his health care provider can refer him for a genetic consultation. The geneticist and genetic counselor will examine your child and discuss your family history with you in detail, asking about any health conditions and/or learning problems in members of your extended family.

The geneticist may also recommend other testing to determine if there is another genetic explanation for your child’s delay. These other tests may include chromosome testing and testing for fragile X syndrome. If these tests are normal but your family history is suggestive for XLID, the geneticist or genetic counselor may talk with you about ordering genetic testing for XLID.

While it is not possible to cure XLID, early educational interventions and therapies can help your child maximize his or her potential.

**RESOURCES**

Genetic Alliance: www.geneticalliance.org


National Organization for Rare Diseases (NORD): www.rarediseases.org
Emory Genetics Laboratory (EGL) is a comprehensive clinical genetics testing laboratory specializing in molecular cytogenetics, rare disease testing, and newborn screening confirmatory testing.

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