



# Center for Human Genetics, Inc.

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## **DNA DIAGNOSTIC TESTS: AUTISM; AUTISM SPECTRUM DISORDERS; INTELLECTUAL DISABILITY**

### **Mental Retardation/ Developmental Delay**

Panel 1

- 6.0 SNP microarray
- High resolution chromosomes
- Fragile X

Panel 2

- IL1RAPL1
- SYNGAP1
- XLMR Tier A (*NLGN3, NLGN4, MECP2, CDKL5*)
- XLMR Tier 1 (*DLG3, FTSJ1, JARID1C, PHF6, ZNF41*)
- XLMR Tier 2 (*GDI1, FACLA, OPHN1, PQBP1, TM4SF2*)
- XLMR Tier 3 (*ATRX, FGD1, MID1, PLP1, RSK2, SLC6A8*)
- XLMR Tier 4 (*AGTR2, ARHGEF6, MED12, PAK3, SLC16A2*)

### **Mental Retardation + Autism Spectrum Disorder**

Panel 1

- 6.0 SNP microarray
- High resolution chromosomes
- Fragile X

Panel 2

- Angelman/ Angleman-like syndromes (Angelman methylation, *UBE3A, SLC9A6, TCF4*)
- XLMR Tier A (*NLGN3, NLGN4, MECP2, CDKL5*)

### **Mental Retardation + Macrocephaly (OFC >2SD)**

- 6.0 SNP microarray
- High resolution chromosomes
- Fragile X
- PTEN*
- RAB39B*

### **Mental Retardation + Epilepsy**

Panel 1

- 6.0 SNP microarray
- High resolution chromosomes
- ARX*
- SCN1A*
- RAB39B*
- Epilepsy Panel 1 (*SLC9A6, PCDH19* (females only), *MECP2, CDKL5*)
- Epilepsy Panel 2 (*ATP6AP2, SLC6A8, OPHN1, SYN1*)

Panel 2

- SYNGAP 1
- MELAS
- Angelman/ Angleman-like syndromes (Angelman methylation, *UBE3A, SLC9A6, TCF4*)

### **Autism Spectrum Disorder**

Panel 1

- 6.0 SNP microarray
- High resolution chromosomes
- Fragile X
- MECP2*
- NLGN3, NLGN4*

Panel 2

- SYNGAP1
- PTCHD1
- GDI1*

### **Autism Spectrum Disorder + Macrocephaly (OFC >2SD)**

Panel 1

- 6.0 SNP microarray
- High resolution chromosomes
- Fragile X
- PTEN*

Panel 2

- NSD1* (Sotos syndrome)
- RAB39B*

### **Mental Retardation (Nonverbal) OR Autism Spectrum Disorder + Microcephaly (OFC <2SD)**

- 6.0 SNP microarray
- High resolution chromosomes
- Angelman/ Angleman-like syndrome panel (Angelman methylation, *UBE3A, SLC9A6, TCF4, ZEB2*)
- Rett (Classic, Atypical, Congenital variants)/ Rett-like syndromes panel (*MECP2, CDKL5, FOXG1, TCF4*)

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