CORIELL INSTITUTE FOR MEDICAL RESEARCH

NIGMS HUMAN GENETIC CELL REPOSITORY

GENERAL CLINICAL DATA ELEMENTS FORM

Sample ID#: _____ **Diagnosis:** _____

Age at Diagnosis: _____ **Age at onset of symptoms**: _____

Diagnosed by: ☐ Specialist: _____

☐ Pediatrician ☐ Geneticist ☐ Primary Care Physician
☐ Maternal Fetal Medicine/Genetic Counselor (prenatal testing)

Phenotype *(please describe dysmorphology, behaviors and other clinical features in support of diagnosis*):

Assistive devices: ☐ None ☐ Wheelchair ☐ Braces ☐ Orthotics ☐ Hearing aid ☐ Service animal
☐ Communication/learning device ☐ Other: _____

Cytogenetic Testing *(please attach a copy of results if available):*
Karyotype (*current ISCN nomenclature*): _____

Test Methodology (FISH, aCGH, etc): _____

Molecular Genetic Testing *(please attach a copy of results if available):*

Gene(s) Tested: _____
Allele 1 _____ Allele 2 _____
Test Methodology (PCR, Southern Blot, Sequencing, etc.): _____

Biochemical Testing *(please attach a copy of results if available):*

Enzyme(s)/Activity Level(s): _____

Other Testing (imaging, EKG, EEG, biopsy, pathology, etc.) *(please attach copies of test results):*

Test/Result: _____
Test/Result: _____
Test/Result: _____

Treatment and Management (*check all that apply*): ☐ Physical therapy ☐ Occupational therapy
☐ Psychological therapy ☐ Speech language therapy
☐ Medication(s): _____
☐ Surgeries: _____
☐ Other: _____
Please add or attach any other relevant information:
_____