

## GENETIC DISEASES EVALUATION CENTER

duzengenetics.com • duzen.com.tr

### Test Name & Code:

- **Test Name:** *GCH1* Sequencing
- **Test Code:** 3872

### Related Diseases & Genes:

- “DYSTONIA, DOPA-RESPONSIVE; DRD” (OMIM #: 128230) || Gene: *GCH1* (OMIM #: 600225)
- “Hyperphenylalaninemia, BH4-deficient, B” (OMIM #: 233910)

### Synonyms:

- GTP CYCLOHYDROLASE I
- DYSTONIA 5; DYT5
- “DYSTONIA, PROGRESSIVE, WITH DIURNAL VARIATION”
- DYSTONIA-PARKINSONISM WITH DIURNAL FLUCTUATION
- “SEGAWA SYNDROME, AUTOSOMAL DOMINANT”
- “DYSTONIA, DOPA-RESPONSIVE, AUTOSOMAL DOMINANT”

### Test Details:

- **Methodology:** DNA Sequencing
- **Performed:** Everyday
- **Reported:** 4 weeks

### Specimen Details:

- **Specimen Collection:** Peripheral blood
- **Container:** Tube with EDTA
- **Specimen Volume/Amount:** > 5 ml
- **Storage/Transport Condition:** Refrigerated
- **Unacceptable Specimens/Conditions:** Serum; Frozen specimens; Hemolyzed specimens; Icterus; Lipemia

### Testing Area(s):

- Dysmorphology
- Neurogenetics

