

Copper Toxicosis

Our DNA assay for this inherited form of Copper Toxicosis detects the mutations in these two genes:

ATP7A: a modifier mutation that can reduce the effects of the disease mutation. Females may have two copies (double positive), one copy (positive) or no copies (negative). Males may have one copy (positive) or no copies (negative).

ATP7B: the disease mutation. Dogs with one copy or two copies of this mutation are both considered at risk for developing the disease.

The DNA test for disease mutation *ATP7B* reveals one of 3 possible genotypes:

- **CLEAR** (those having 2 copies of the normal allele and appear to be normal).
- **CARRIER/AT RISK** (those having 1 copy of the normal allele and 1 copy of the disease mutation = 1+ At Risk for clinical signs.).
- **AFFECTED** (those having 2 copies of the disease mutation = 2+ At Risk for clinical signs.)

| <i>ATP7B Disease Mutation</i> | CLEAR Male | CARRIER/AT RISK Male | AFFECTED Male |
|--------------------------------------|--------------------------------|---|------------------------------------|
| CLEAR Female | 100% Clear | 50% Carrier/AtRisk 50% Clear | 100% Carrier/AtRisk |
| CARRIER/AT RISK Female | 50% Carrier/AtRisk 50%Clear | 25% Clear 50% Carrier/AtRisk 25% Affected | 50% Carrier/AtRisk 50% Affected |
| AFFECTED Female | 100% Carrier/AtRisk | 50% Carrier/AtRisk 50% Affected | 100% Affected |

The protective mutation *ATP7A* works to provide some form of risk reduction from the disease mutation, when present. The number of possible copies is dictated by gender. Only the X chromosome can carry a copy of the *ATP7A*, which is why males cannot carry two copies (they are XY). The Y chromosome will not carry a copy of the protective mutation.

| <i>ATP7A Protective Gene</i> | NEGATIVE Male | POSITIVE Male |
|-------------------------------------|---------------|---------------|
| NEGATIVE Female | 0 Copies | 1 Copy |
| POSITIVE Female (Carrier) | 0 or 1 Copy | 0 or 1 Copy |
| DOUBLE POSITIVE Female | 1 Copy | 2 Copies |

The effects of the disease mutation may be reduced based on the dog's genotype for the protective mutation.