SB0188: Public Health/Rare Disease Advisory Council, 2/7/23
Jeneva Stone, Rare Disease Caregiver (Written Testimony)

My son Rob Stone, 25 years old, an artist and advocate, has an ultra-rare disease, dystonia 16. He's the only reported case in the U.S., among only 20 cases worldwide. Dystonia 16 is so rare that we spent 14 years searching for a diagnosis that is still only available through genome sequencing—a test that analyzes the more than 20,000 protein-encoding genes in DNA.

Dystonia affects muscle contraction. Our muscles work in pairs: one contracts (flexes) and the other relaxes (extends). When someone with dystonia tries to initiative movement, both flexor and extensor muscles tense up at the same time. Dystonia can be focal (affecting only certain parts of the body), or global (affecting the entire body). It can be painful—writer's cramp is a mild, incidental form of dystonia. There are dozens of dystonias, linked either to a genetic mutation or to defined clinical symptoms.

As a result of his rare disease, Rob also has disabilities, and while Maryland has strong supports for disabled persons, the needs of the rare community do not fully align with the disability community. Our family struggles with prior authorization for life-saving medications, with coverage for the high-dose supplements research has shown are essential to his well-being, with continuity of care, and with access to Medicaid services.

Rob's throat muscles are affected, and he has life-threatening gastroesophageal reflux disease (GERD). He cannot swallow, and needs an omeprazole solution that can go through his G-tube. Keeping this prescription approved is an ongoing Medicaid nightmare. Research is being done on Rob's cells, and we have discovered that high-dose biotin, thiamine, and luteolin (vitamins and a flavonoid) stabilize him and appear to be preventing his disease from progressing. Luteolin slows and stops cell apoptosis, or cell death. Maryland Medicaid constantly erects barriers to these therapies in the form of bureaucratic harassment.

Services are also a problem. Rob was not enrolled in the Rare and Expensive Case Management program (REM) until he was 15 due to wait lists and lack of a diagnosis. There are 302 qualifying diagnoses for REM, yet there are 7,000 rare diseases—REM serves less than 1 % of the rare disease population. Furthermore, of those qualifying diagnoses, 68% end at the age of 21. Rare adults are often left out in the cold.

While I applaud Maryland's progress with newborn screening, it would not have diagnosed Rob. Nor would it my dad and sister, diagnosed with rare diseases at the ages of 70 and 40, or my hair stylist's mother, diagnosed at 60.

Rare Marylanders need access to genetic sequencing, adult services, experimental treatments and fair insurance guidelines. We need a Rare Disease Advisory Council so we may have a voice in shaping the policies that impact our lives.