Box 5: Disease Severity

[5a] Parent participant (10004): "*He had almost a compete deletion in his chromosome. Anything could just set it off…They were like 'It can go off anytime. It can cause another hyperammonemic episode… He could possibly die, or go into a coma, or have seizures…This is his best chance of survival.' That's what we did….it was really the only long-term option for him.*"

[5b] Parent participant (10006): *"[He] had 0% of the OTC enzyme. Honestly, we really weren't given a choice. It was sort of a no-brainer like, okay, you live in fear for the rest of your life that when he gets sick his ammonia will go up and cause further brain injury. That's what we chose really with the guidance of the metabolic team."*

[5c] Parent participant (10023): "*I don't really know. I feel like she's probably always going to be a little bit unstable. We seem to be in and out of the hospital every three months, about. I don't know. She has zero enzyme function, so it's pretty black and white in her case…. I feel like things probably aren't going to change very much, but who knows?... Do we do it now and protect her neurological development, but then risk her life, or do we continue with conservative management in the hope that maybe an alternative treatment's going to develop?*"

[5d] Provider participant (20006): *"I think in the severe neonatal onsets; I think that's less of a question at this point. That's really the only way to save them….I think that in that case, there's…less question about whether it's the right thing to do…In the later onsets, where it's a little bit less clear-cut, I think – we have extensive conversations."*