

Box 4: Tipping Point

[4a] Provider participant (20010): *“Finally, push came to shove where it was the kids were coming in too frequently, or their ammonias were being too problematic, difficult to treat. Then we finally made the decision when that balance or the scale seemed to tip... Every child is different, and every child comes in with a different set of issues. Whether it’s the diet, whether it’s the medicine, whether it’s just the lack of activity or just feeling out of it all the time, whether it’s hyperammonemic crises, bringing them in frequently, these are all issues that come into the play of whether or not you go ahead and you finally make a transplant decision.”*

[4b] Provider participant (20011): *“Families take that information in different ways. I’ll have kids that I think have a little bit milder disorder, where we’ve talked about transplants because I usually do in the urea cycle defects. I at least let them know what the spectrum of options are...The family, well, when can we do the transplant? I say, your kids aren’t that sick. Then we have other ones where the kid’s coming in once a month, and they’ve had many [hyperammonemic episodes] ...and the families say, oh, I don’t know if I want a transplant.”*

[4c] Provider participant (20012): *“For some caregivers it’s right away. They want the transplant right away. They want this condition gone. This condition scares them way more than a transplant. There are some caregivers who, within a year, would say, “I want a transplant.” ... Some families just reach it right away and others have to sit for a while with it.”*

[4d] Parent participant (10026): *“It was his second crisis then at 10 months, I guess,...that tipped the scales.... After the second crisis, there was – we didn’t feel like it was a – it was a decision that was definitely transplant.”*

[4e] Parent participant (10002): *“Honestly, as caregivers we were not handling it well. We were handling it but to think of living years like that, with that kind of fear, was overwhelming.... I think for us it was the fear of her just having a completely severe decompensation to the point where she’d have a really severe brain injury and that her life could completely change in an instant... I think it’s one thing that if your baby’s born a certain way and that’s what it is and you deal with that situation. When you’re just constantly facing it but knowing that you’re not there but that everything could change for them at any time, to me it’s just a really different way to try to exist with just the fear of it.”*