

Box 11: Access to Quality Metabolic Care

[11a] Parent participant (10008): *“Yeah, the quality of care. Part of my decision [to transplant] too was UCD being so rare. What I was told is I can’t be on vacation and be sure to find somebody that’s even going to know what I’m talking about. Even our local hospital didn’t even – they had no idea what a UCD was. They had no idea how to care for him. I just felt it was really dangerous for him to live with that disorder in the future, for his life.... I didn’t feel like I was getting much help with the nutrition aspect, and I was worried about that for the future because it’s such a huge part of keeping him healthy... Then a couple of times...he had an emergency. He had been vomiting all day, and we took him to the ER. Of course, the ER was terrible. They don’t know anything about urea cycles.”*

[11b] Parent participant (10026): *“All these little things were adding up for us. Okay, he didn’t have a crisis for eight months, but they’re not able to quickly handle it. This is a hospital that actually knows about it. I had talked to plenty of caregivers whose child had brain damage because they were in a place...where the hospital didn’t know about it. Ours knew, and they still took days to get this going and get him stabilized....we just looked at each other and said, if we weren’t here watching like hawks all the time, would he survive?...When we went in there, my faith was that these people were going to save his life, and suddenly, I couldn’t count on them to do it right. You know?... The head of transplant came in, and said again, ‘I think you need to talk to us again.’ We said, yes, we wanted to get him – we changed our minds. We want a transplant.”*

[11c] Parent participant (10012): *“We’re here...with very limited access to a decent metabolic geneticist. I’ve been told there just aren’t that many, I guess. There certainly aren’t any in this state, and so it’s just access to good care, basically...It seems that there are only a handful of specialists throughout the country, and if you’re not in that location, you’re really subject to pretty subpar care.... Not being able to rely on your healthcare provider. I mean, I think the question had to do with local – not having local access to good physicians. We never felt like they had our backs here. We could never rely on them. We would put a call in and not – we wouldn’t get a call back for a week. When you’re trying to manage day-to-day, it’s like, oh, we got these labs back. What do we do now? You can’t wait a week. I mean, it was horrible, so that was a huge stress for me knowing that we were basically on our own.”*

[11d] Provider participant (20021): *“If you have a medically unstable child who lives a long way from a facility with any special expertise with urea cycle disorders, he could get into trouble quite easily and not have medication at hand that they need. That obviously does change the balance of how you would recommend liver transplant... I have a [adolescent] patient who – she was really well her whole life, but then...had one or two hyperammonemic episodes, not even that severe, quite moderate. Because of her geographic location, I recommended transplant because her ammonia control baseline wasn’t that good. I was worried about compliance, and I was worried about her geographic location, so we listed her, and she was transplanted last year.”*

[11e] Parent participant (10028): *“I’m confident that if she does get sick that we’re in the right place. I trust the team here very much... Here, anytime there’s been a question of high ammonia, they’re like come in. Let’s do labs. We wait in the hospital until we know what the number is...The reason [liver transplant is] not forefront in my mind is because we are with [this] department, and we’re with [this] doctor. I’m very confident in our team on our day-to-day management and how we do everything...If it’s decided that we can’t stay here... I pretty much know transplant would happen in one of two facilities, and then we’d go from there.”*