

Box 12: Metabolic Physician Approach to Treatment and Guidance

[12a] Parent participant (10027): *“I had such an incredible team that I really didn’t have to navigate it alone. They were really there for me...The doctors have been incredible...I never really had to navigate those things alone medically.”*

[12b] Provider participant (20001): *“I think [the family’s] view of transplant is impacted significantly by the impression they’re getting from both the metabolic genetic team, who usually know them really well and is really involved with them, and the transplant team that they see. From what I’ve heard from other doctors and from families, they may get a very different impression of what transplant involves and what the long-term outcomes are depending on who they talk to.”*

[12c] Provider participant (20001): *“It seems like the discussions I’ve had with geneticists is that people have very different approaches at different institutions.... Some people have very clear ideas about who they refer for transplant, and some people don’t. I don’t think we know which one is the right approach...I was definitely surprised about that when I started talking more to metabolic geneticists at other centers or that had recently come from another center to ours.... I think it has a lot to do with, especially if you only have a few cases and then you have even fewer cases who decide to go through transplant, what happens to them afterwards. If you see a bad outcome or two that can totally change your impression for the next 20 years versus if you see some who do really, really great, then that may also change your referral pattern.”*

[12d] Parent participant (10011): *“He was very, very anti transplant, very anti transplant, just really had nothing positive to say about it, and he remained that way. I will say that’s part of the reason [my son] was so much older; why we didn’t give it more consideration...There were a few periods where he was really sick for a really long time and almost died a couple times, and still, this doctor held fast. He did not think transplant was the way to go...When I’d bring it up to the doctor, he still was not in favor of it, so we just didn’t really push.... I still was not positive it was the right – at that point, I still was a little unsure. I mean, having a doctor for 20 years that is the top of the field saying don’t do it. Don’t do it. It’s like, oh, man, are we asking for trouble doing this?”*

[12e] Parent participant (10019): *“I would say, for our family, one of the biggest challenges was that we didn’t have support from our genetic team... They weren’t encouraging a transplant. [The hospital] believes that you’re much better to manage the disorder. Because we trusted these people because they saved her life and because they kept her alive, we had our full trust in them with how to handle her. Then when we decided to look into transplant, and it wasn’t even something that we could bounce ideas off of them; it was something we just didn’t talk about. I would say that was, for us, one of the bigger challenges.”*

[12f] Parent participant (10012): *“We mostly spoke with the geneticist...and if you’re familiar with him at all, he is of the mindset that if you’re born with OTC, basically, you need a transplant. With that mindset, it just – it was like this is happening. This is the best solution. Oh, yeah. By the way, there are risks, and here is what they are. It wasn’t even like a risk benefit analysis so much. It was just be prepared. These are the things that could happen, full well knowing that this is going to be a far better choice for your daughter at this stage. That was already the foregone conclusion, I think, when we arrived.”*

[12g] Parent participant (10031): *“We did pursue transplant. [Our doctor] has just been open to what we as caregivers feel comfortable doing. He thought it was important for us to explore and see what would be beneficial for [our son].”*

[12h] Parent participant (10023): *“It just seems like – because there’s no standard practice in these cases, it’s extremely difficult. It feels like we’re on the frontier of this thing, and it’s not clear what the right decision is, and nobody really wants to say one way or the other.”*

[12i] Parent participant (10008): *“I mean, if I could change it, I really wanted to have more guidance from [my son’s] metabolic doctor, but he never really gave much of an opinion.”*

[12j] Provider participant (20006): *“What we often tell the family is that there’s not a right answer here. It’s not wrong to choose one or the other. It’s what’s right for them as a family together. That, I think, is difficult for families because in something so major, I think what they really want to hear often is, ‘This is what you should do. This is what you need to do for your child.’ Making that decision and putting it on a family is really difficult. I always feel that when we have these discussions that I wish I could just tell them what to do. I think that would take a lot of the burden off the family.”*