Supplementary Table: Additional Illustrative Quotes

|  |  |
| --- | --- |
| Theme | Additional Quotes |
| Receiving Diagnosis Symptomatically | |
| Parents experience significant delays in diagnosis despite persistent advocacy | So we even went to a genetic doctor…And he saw [child] and she had a really good, she was a chubby baby. She had a good muscle tone and everything. And he asked us, like, “why do you bring her here?... this baby's totally fine”. (Symptomatic, Mother)  So I did take her to the doctor numerous times. Pediatrician and a local children's hospital. And they always said there was nothing wrong with her. (Symptomatic, Mother)  When she was three months I, I made an appointment and took her to her pediatrician. And the pediatrician…she just kept saying, like, I can't really call it anything yet. She's just a little twin…She's like the underdeveloped twin. She kept telling me to, like, look at her more, hold her more, like, pay more attention to her. (Symptomatic, Mother)  I realized that her tongue was like shaking a lot…so I asked the doctors about that and once again, they just said, our local hospital said, you know, she's premature. She's just, you know, developmental delays. (Symptomatic, Mother) |
| Receiving Diagnosis on Newborn Screening | |
| Parents have limited knowledge of NBS prior to child’s diagnosis | I knew very little about the newborn screening knew that when he was born, they took him back to the nursery and did a heel stick. And I was really unaware of all of the things that they were screening for…to be honest, I didn't even know SMA was on that screening. (NBS, Mother)  So we knew absolutely nothing about newborn screening. I knew that there were blood tests that they did in the hospital when a baby's born, but I did not know anything about SMA, I didn't know that was part of it at all. (NBS, Mother) |
| NBS disclosure leaves parents wanting more information, highlights need for current information | So if I was telling somebody about their kid, I probably would've went into a little bit more detail about it only because, you know, that shocking news...Like there would have been treatment, like how the treatment would work. (NBS, Mother)  They said your baby will lose her muscle, like all her muscle, so like wouldn't be able to like eat, talk, move, sit up, would need to be in a wheelchair, could possibly be on a ventilator, and that's pretty much was how we took it. And then they said that there is treatment, but they didn't give no information on treatment at that point. (NBS, Mother)  Or at least giving…some sort of idea as to what to expect...'Cause if they would have at least told me the diagnosis, it would have gave me that peace of mind. (NBS, Mother)  I wish they would have told me what it was instead of keeping it a secret, because that way is…less stress on you than to like, be worried about what it is that they have, what's wrong and everything like that. Because it just makes you ten times more paranoid and worried not knowing it at all. (NBS, Mother) |
| Diagnosis in ‘healthy’ child leads to disbelief and denial in parents | And at that point, we were pretty much still in denial. I mean, that was kind of like when our world came crashing down…Our son was extremely strong. He was not showing any weakness, at least in our eyes. We had no idea…every time we took him to the doctor, he kept saying, you know, going on and on about how strong he was. And then they were telling us that he had this disease that was going to affect that. (NBS, Mother)  At that time like, I was still second-guessing if it was as serious as everyone was saying it was because, like, my son never really showed any development issues, never showed any of the signs that I googled. (NBS, Mother)  Yeah, I was like, there's no way. Like are we really talking about the same kid…look at him. (NBS, Father) |
| Parents supportive of NBS for SMA, recognizing need for early diagnosis | I think it's just so important, especially for SMA, because it can change it so much. And I think as hard as it is to learn that information when you've just had a baby…it's still better than learning it down the road…I think it'd be harder if I had thought everything was fine for months, and then finally found out something was wrong. I think- I prefer to know up front, and just find out then and start working as soon as possible on what we can do about it. (NBS, Mother)  No, no mixed feelings. No, I'm 100 percent grateful for the newborn screening. I, I give that, the entire credit to pretty much saving our son's life. It gave him a life, a whole different life. It gave us the opportunity to receive treatment. And we don't know when he would have started showing signs because he does have four copies. And so that to us is just, we are so, so grateful for the newborn screening. (NBS, Mother)  And you know, there's a part of me that's grateful that, you know, we live in Ohio and it was caught he was first born, so he was able to get treatment early and therefore will probably go on to live a nice, long healthy life, because I know not everybody is that lucky. (NBS, Mother)  Yeah, that's, that's a game changer. That's amazing. You know, I definitely recommend it...just 100 percent to do it, to get ahead of it. Because the sooner you do it, the the better your kid will turn out, you know, and have a chance at a more normal life, better outcomes and everything. So definitely 100 percent support it. (Symptomatic, Father)  I don't know why it's not in all 50 states by this point. We know families that were not diagnosed until six months of age or longer and their outlook is a lot different than ours, and there's things that our son can do that were never dreamed of before treatment and...time is, is the number one thing to get ahead of SMA, the earlier the better…So I think that's, it needs to be in all states because time is of the essence. (Symptomatic, Father)  If I could change it, I would definitely have the test for SMA be on the newborn screening when my daughter was born, that would have been a much faster and effective way to diagnose her. (Symptomatic, Mother) |
| Parents of children diagnosed symptomatically left wondering how their children’s lives may have been different with NBS | She was born in West Virginia, and my understanding is that they started newborn screening for SMA two months after she was born. So that brings us to the fact that if she would have been born on time and not premature, then she would have been newborn screened for it. And this whole situation probably would have been much different…Every state should newborn screen for it, because that's you know, you're going to get your highest chance of an effective treatment at newborn versus months later when symptoms have already kicked in. (Symptomatic, Mother)  It's amazing really, because I am honestly bitter about it, because [child] just missed it but for future kids it's great. (Symptomatic, Mother)  I obviously still have my days where I'm like, this sucks, I wish this never would have happened and...for the first year after diagnosis, I just kept wishing we would have caught it sooner because he, he didn't show any symptoms for so long that I was like, man that first time I saw that hand tremor, if we were to caught it right then, he would've been treated right then. He could be walking. (Symptomatic, Mother) |
| Counseling for Parents with Children Diagnosed on NBS | |
| First appointment overwhelming for newly diagnosed families | Honestly, I don't, I don't really remember. It's like a blur. And I that upsets me because, like, that's something I should vaguely remember. But I was so scared and not understanding anything they said (NBS, Mother)  Just the nature of like, it's hard to absorb information at that stage when you're so- it's so new and you're so- your world has been completely rocked…But that wasn't because of how they did the meeting, it's just hard for us to absorb at that stage when you've not fully accepted or come to terms with what is happening. (NBS, Mother)  It felt like it was everything was going really fast. The information I was being told, because like, it was shocking that she had it, and I was like scared and everything. (NBS, Mother)  But you know, I'm also in a haze and I, you know, nursing my son and in between doctors coming in to tell me about what's wrong with him. But everybody was really nice and they were very patient and went over stuff if there was something we didn't understand necessarily, but I also remember being just in shock basically. And you know, at that point, you don't even know what questions to ask, necessarily (NBS, Mother) |
| Provider compassion key for parents in initial SMA appointment | No, she was very thorough and very, a lot more sympathetic…they definitely took their time to, like, realize that we're parents of a newborn baby who just got diagnosed with the worst, this awful condition. And, you know, we were emotional. It was an emotional thing. And they were definitely a lot more caring and compassionate. (NBS, Mother)  Everybody has been super supportive and helpful…with all the people that are seen and everything that's going on there and stuff, it seems like we were treated special. Like we never felt like we were just another person there and like everybody seemed to go out of their way to help us and make sure we knew what was going on and understood everything and were comfortable. (NBS, Mother)  I thought everybody handled like handled us really well because we were both, I mean we're new parents…I look back and like now I'm like thankful for the way everything went down and everybody we've seen because they've all been so nice to us. Even when we go to [hospital] now, they're like, I don't know, it's just nice to see that they like appreciate him, and know him, and want to see him, if that makes sense. (NBS, Mother)  I think the people we met with were all very gentle in talking to us, and that was good. It was like they were sensitive to how shaken up we all were. And I think they explained it very well. They took their time and drew it out on paper for us and made sure we really understood what the disorder was, and then let us ask all the questions about the treatments. (NBS, Mother)  Right, she didn't leave anything unturned. She was very open and honest and it made it so good because we knew what to expect. We knew what to look for if something were to start going wrong (Symptomatic, Mother)  They were just, they were upfront and realistic, basically. But very sympathetic at the same time. It came off better hearing it from her than it had anybody prior. (Symptomatic, Mother) |
| Treatment with Gene Replacement Therapy | |
| Gene Replacement Therapy Parents’ First Choice for Treatment | I remember thinking, you know, well gene therapy sounds like it would be the best option…being able to do one treatment and be done is much easier for everybody involved…it will be far less traumatic for my son to get a treatment when he's a baby than to have to get shots for the rest of his life or take a pill every day for the rest of his life. (NBS, Mother)  No, we looked at it as the best thing that could ever happen to us if we could get that. So we were hoping and wishing for that from as soon as we found out about it. We- yeah, we were not at all concerned about- we only looked at it as a positive. We even said that we would rather get the gene therapy than win the lottery. I mean, it was like, that's what we would pick. So that was like our saving grace. (NBS, Mother)  My thoughts were that one of them was to be administered every single three months for the rest of their lives and the other would be administered just once in their lifetime. That's what I remember. (Symptomatic, Father)  We both kind of agreed that we didn't want to look back in a year or two years, five years and think, you know what? What could, how strong could he be if we would have chosen gene therapy over Spinraza? You know, what, he's gained a lot with Spinraza, but could he have gained more with gene therapy? So, we just went ahead and did it and were thankful that we did. (Symptomatic, Mother) |
| Parents seeking gene replacement therapy for symptomatic children race against disease progression | We were just kind of desperate…we were willing to try anything, just, you know, so you wouldn't lose more functions and maybe she could recover and get a little better because she was tired…that's when she lost her swallow...she started deteriorating more. She was losing energy. She's more lethargic and couldn't, you know, keep her neck up. And so we were just, we were, we were you know, we were willing to try anything. (Symptomatic, Father)  I think we still would have went for it, regardless of our feelings…because at this point it was life or death…it was our only hope at the time. (Symptomatic, Mother)  But I mean, we were basically just handing over our child for an experiment and, but like I said, we, we discussed all our options and it just felt like the best thing to do and it was a chance. (Symptomatic, Mother) |
| Parents with children diagnosed on NBS anxiously await signs that gene replacement therapy worked | The only concern I had and I still do, I still have a concern is like, how do we know it's working? How do we know that his body accepted it…But I'm still I still have the uncertainty to me, like, how do we know that it's working? (NBS, Mother)  Well the thing is, I treated my child with something that I was told worked…but I have never seen the need to even treat him, I've never seen it: he was symptom-free before he was treated. (NBS, Mother)  But like for me to look at her now, I still don't see nothing wrong, like, from the time that I got this information I thought she was able to move her arms, move everything. So, then we did the treatment and she's still the same way. (NBS, Mother) |
| Parents with symptomatic children able to see improvements more quickly | I would like to say that I honestly, I don't even, I'm not concerned because gene therapy gave my son literally the ability to eat. He eats table foods now and drinks by mouth and doesn't require anything through his G tube. And I cannot explain how huge and life changing that is…He remembers food, he remember those things. It wasn't like he never did eat. And we couldn't eat dinner together. We couldn't eat around [Child] because it was just torturous…no one had told me that that was even going to be a possibility...there's no price for that. (Symptomatic, Mother)  Every time I post about anything that he's gained or any of his strength that he's getting, or gaining back or anything, I'm always like, we are so, so, so thankful for this gene therapy. Like, I fully believe that if he did not get gene therapy, he wouldn't be where he is today. (Symptomatic, Mother) |
| Impact of Diagnosis on Parent and Outlook on Future | |
| Parental anxiety improves with time, but does not disappear as uncertainty in future remains | I guess early in the beginning, every day I was waking up looking for symptoms, kind of examining her to make sure that she was fine. Now, it's been long enough, I kind of take it for granted…A lot of times I think I forget about it, which is kind of weird, but- I guess it has changed, because in the beginning, you know, I was appreciating every day more and thinking, you know, I don't know if she's going to live to be two…And now it's, I see myself planning the future more, so it's definitely a good thing. (NBS, Mother)  Well, I'm not emotional anymore. I'm not just randomly freaking out. I'm not basically being a mom-studier, if that makes sense. When I first found out I would like observe my son an awful lot, and now it's like, OK, I'm no longer basically studying him. Now I interact more with him, and I don't know. It's changed for the better. (NBS, Mother)  I'd say it's way better?...But, you know, we were still really depressed and we weren't as hopeful until she did get genetic therapy. But since she's had the therapy and we've seen all these improvements, I mean, it's, it's a lot better. It's better for her, is better for us, for our mental health and everything. So it's a huge, huge difference. And we're just grateful and happy and we thought-- we feel like we won the lottery, basically. (Symptomatic, Father)  I think that's the thing that most parents struggle with, probably because the future is so unknown. You know, you don't know what potential regression might look like. You don't know what symptoms might occur. Maybe they don't occur when they do. Like you know, how old is he? What is the effect on him? (NBS, Father)  I still have the uncertainty to me, like, how do we know that it's working? How do we know that it's, you know, something that's good for him, that it's going to help him in the future? And if it is, how long? Is this a one-time thing or will he have to do it again? So I still have those uncertainties that I have never got like a straight answer with. (NBS, Mother)  So that is, I think the most difficult part in the whole thing is, is it working? Is this just him?...Is it going to last forever? You know, those are kind of the questions that we have and the feelings that we have about it. It's just something that we will always question if we see anything...I know it's not considered a cure, but it this going to keep him from not ever showing signs because he's never shown any signs yet. So those are just kind of things that we ask a question pretty much on a daily basis. (NBS, Mother) |
| Parents remain cautiously optimistic for children’s future | I think I'm more optimistic today, I think seeing him meet milestones really helped with that. The first time he rolled over, I had a huge sense of relief, I was like, "oh my gosh, he's moving forward, he's not just stagnant. Something's happening, something's working." I think that was reassuring….So I think I'm more optimistic now, having had the treatments and stuff, than at the beginning. (NBS, Mother)  It's not only stopped the progression, it seems, but she was able to regain some of her strength back that they did not expect. Her swallow came back 100 percent…it took her a few years, but then she was able to sit up unassisted on her own and just seeing all those milestones met, gave us a lot of hope for the future. And so now, we very rarely think of the negative or like what will we have to do if this happens, you know, we're, we're so much more hopeful for the future (Symptomatic, Mother) |