You are listening to a Rare Genomics RareShare patient navigation podcast. We are an organization and Community dedicated to helping and supporting people with rare diseases.

Imogen: Hi everyone. My name is Imogen Crispe and I'm your host for today's podcast. I'm a volunteer with the Rare Genomics Institute and RareShare.

The rareshare.org social networking platform provides an online community and support system for rare disease patients, families, and health care professionals who may otherwise not know anyone else suffering from their disease. Just a reminder that this podcast content is for general informational purposes only, and in no way should it be considered a substitute for medical advice, or for personal diagnostic, or treatment purposes. Now, being born with and growing up with a rare disease means you don't necessarily know any other way of life.

Today, I'm speaking with twenty three-year-old Anna Laurent who was born with Alagille Syndrome and as she grew up, she gradually learned about what that meant and how her life was different from other kids' lives. She is going to tell us about her symptoms and treatment experiences, participating in research, and her involvement with rare disease advocacy. Anna also recently graduated from college so she will tell us all about her new job. So thank you so much for joining us today Anna.

Anna: Thank you so much for having me.

Imogen: So maybe we can start and you can tell me about how and when you found out about, and actually understood what Alagille Syndrome was.

Anna: I was diagnosed when I was about 4 months old and at that point very very little was really known about Alagille Syndrome. So it took my parents going to about three hospitals and four different states to really find a diagnosis. Of course, I don't remember any of that because I was a baby. But to be honest, I really didn't understand that I was different really until elementary school when my peers, so kindly, pointed out to me and

that was because I was very short. I still am, but I was really short when I was younger. When it comes to actually understanding it, that was a journey. It really began in middle school, just had [begun] trying to understand what it was exactly that I had but I really didn't begin to really truly understand the complexities of the condition until I attended my first Alagille Syndrome symposium as a young adult, right before starting my first year of college.

Imogen: Yeah. I was thinking you probably wouldn't have fully understood as such a young child. That's really fascinating how you can gradually learn more about it. And for people who don't know much about Alagille Syndrome, can you tell us about the signs and symptoms?

Anna: Of course. So Alagille Syndrome is a rare genetic disorder. It affects roughly 1 in 70,000 people. It's very complex in that it can really affect multiple major organ systems with a wide range of severity and there's no cure at this time. For our warriors, the most common, as common as anything can be in the rare disease world, is the first sign being jaundice upon birth and cholestasis, which is the blockage of bile flow from the liver. So that's when they have liver issues and as they get older, a lot of times they also begin dealing with a severe chronic itch which involves the liver. But that's just the liver side of things. We also have many warriors that deal with heart issues as well, just like a heart murmur, congenital heart defects, and things like that. But like I said, the severity really ranges a lot. So you have some warriors that don't have any issues with their liver and it's just their heart that severely affected, or others have kidney and heart issues. So it can be really complex. Each warrior has a very unique journey that they experience a different battle which keeps things interesting. That's a nice way of putting it. I could go on but it's really complex and can be a little bit overwhelming just due to all the different ways it can affect our warriors, but I would like to say for anyone who might be interested in finding out more and really deep diving into it, I highly recommend going to alagille.org. You can find a lot of in-depth information that's in Lay terms. So it's actually easy to understand but that was a good synopsis of it as a whole.

Imogen: Thank you for that overview. That was great. And that's really useful that you gave that link for people. Thank you for that. You already mentioned a little bit about the kind of reaction you had from your classmates. But other than that, what other sort of impacts did Alagille Syndrome have on you as you were growing up?

Anna: I, overall, have been incredibly lucky in my journey with ALGS. My particular case is pretty mild, especially compared to those who need multiple organ transplants and surgery as infants and children. Luckily, I have never, knock on wood, had to have a transplant of any kind or really any major surgeries. But my main issue growing up was my liver and poor weight gain. My parents really dealt with the hardest part of my battle when I was a baby. I was failure to thrive. That was the most life-threatening part of my journey. There was definitely a time where they weren't sure that I was going to make it to the next week, month, or year. But obviously, I made it and I stuck around. But to be honest, I really felt like a normal kid for a really long time other than itching and being short. A lot of that had to do with my parents and my siblings. They were the main reasons for that. I'll go into that more a little bit later. Middle school was really when things got rough and really when the negative impact came in. Middle school is just a really uncomfortable and awkward time for most kids, but add a rare disease, delayed growth, and a chronic itch and you really kick it up a notch. So, it was a really difficult time. I was really beginning to finally come to terms with this disease that I didn't sign up for and was not happy about having and being surrounded by people who didn't understand what I was going through and so I felt very alone at the time. Now looking back, I know what I was experiencing. I was experiencing depression and it was the first time that my mental health was really impacted by having this disease. It was really one of the hardest times for me even though my physical health was actually pretty good at that time. But I made it through that and things really began to improve. I started to actually realize all the strengths that I was gaining from fighting my battle with Alagille Syndrome. I was finally gaining adaptability; I really don't take no for an answer, especially when it comes to being short and things like that. I think all warriors are really resilient and we have a lot of empathy for those experiencing things that we might not

have experienced ourselves just because that's something that we have to deal with a lot. And it really made me a very determined person. But I think overall, growing up with it was a bit of a roller coaster ride. It has ups and downs for sure. But I really think the impact was both positive and negative as I feel like a lot of things in life are well.

Imogen: Yeah, you've been through a lot and that's interesting to hear about the depression that you went through too. I'm sure that's something that a lot of rare disease patients would go through so it's really great to hear that you got through that. Thanks for sharing that. I was wondering if you could tell me about the sort and frequency of the treatments you've received for your symptoms.

Anna: When I was diagnosed, treatment options were very very limited. Really, they still are. I like to call what my treatments were band aids, if that makes sense. They address one or two of the issues but not the source. My treatments were to increase my intake and absorption of vitamins because my main issue is my liver and to improve my liver function as much as we could. As a young child, I was on very high concentrates of liquid vitamins and that went on for a few years actually. Eventually I got switched to just a fat-soluble chewable vitamin of A, D, and K and I took that every day. Then, I took Actigall which has different names now. I was taking that twice a day and that was for my liver function. That remained about the same until high school; then, I started taking over the counter calcium chews as my vitamin D was low. Then, I got into college and my vitamin D was really low. Now, I just take a high concentration of vitamin D once a week and over-the-counter vitamin. So, I'm really not on a whole lot right now. My liver stabilized when I was in high school, so it wasn't getting any worse and it wasn't getting any better. That's really all I'm on right now. A lot of medication was when I was younger and I was struggling to really grow and not just gain weight, but then maintain that weight. That's really all that I had to take growing up.

Imogen: In the video of your talk that I watched, you mentioned that you had been involved in some research when you were growing up and I was wondering if you could tell us a little bit about that.

Anna: I really started participating in research at a pretty young age. I still don't really know a whole lot of what it was because I was so young. From what I understand, I was giving more blood and my mom explained to me that I only had to get stuck once which is really my main priority, not having to have two sticks. But, the first research I really remember walking through and reading what it actually meant and all those things was about tracking the well-being of patients with chronic illnesses. They were just really excited that they had a patient with a rare chronic illness. That was most of what I participated in. There weren't too many drug trials at all when I was growing up and if there were I didn't qualify for them or I wasn't experiencing the issues that they addressed. More recently though, I'm involved in research looking into Alagille syndrome warriors who don't have either of the two genes that have been identified to cause it and that's really really exciting. That's probably the research I've been the most excited about, ever. That's really a good summary of all the research I've been involved with.

Imogen: That's really fascinating because, you know, we get a lot of questions from rare disease patients who are asking about different research that's going on and how they can take part. So, I was wondering would you recommend other rare disease worries take part in research like you have?

Anna:My view on it is that I recommend doing what's best for you and whatever that might mean. It's going to vary from person to person. Like I said, I hope it was obvious, I get really excited when it comes to research. I think research is a really amazing and empowering thing for anyone to be involved in, especially, when you have a rare disease. I feel like a lot of times it feels like we're just going day to day and so when you get to be involved in something that's bigger than yourself and can help future generations of warriors, it's a feeling that you really can't describe. However, I never say I recommend

everyone to do research. It's so important to look into the research and what it means and what it's for and the possible impact it can have on you. That's been a more recent change in my, not opinion, But in my response to people because in the past couple of years, there has been a drug trial going on that treats the chronic itch often experienced by ALGS warriors. About a year ago, I was asked by a parent of a young warrior if I would have participated in such a trial if it was available. My short answer was I probably wouldn't, and needless to say, that did not receive the best reaction and I had to explain that I know if I were to participate in such a trial and my itching were to decrease or stop altogether which would be amazing, of course, but then that trial were to end and I were to stop receiving that drug, that's unfathomable for me. I can't, that would be torture. I've never lived a day without this itch so I don't know anything different. I cope with it, I deal with it. I adapt with it and the thought of experiencing that normalcy, I guess, and then to have it taken away, it's just something that I know in my personal case and where I am in my life, I couldn't do it. It would not work for me. That's why I always say I recommend doing what's best for you. Obviously, I still love research and I will participate in anything really that's offered to me as long as it fits what's best for me. But I think it's really easy for warriors to put a pressure on themselves that they need to participate in research for future generations. And yes, that's an added bonus but if it's going to negatively impact your well-being and your life, then that's not worth it in my opinion. I think there's always going to be research and drug trials that come up every now and then and you're going to have opportunities. So I think that when warriors are able and willing to participate in research and it's what's right for them, it's such an amazing and empowering thing. But I do think that it should not be taken lightly and you really need to look into it and into who's doing the research, also. I've had some amazing experiences with some amazing researchers who don't use you as just numbers and I didn't realize how empowering that was until I got older. I think just not taking that decision lightly and really looking into it, and keeping yourself in mind, and not just putting a pressure on yourself is really important.

Imogen: That's interesting that you were talking to parents of a young child because my next question was going to be what would you say to parents who are thinking of putting their child in a research study? I'm sure there's some similar considerations that you've just mentioned but is there anything else that you would you would add to that?

Anna: My mom actually handled it so well with me, I feel like. I remember the first time that I participated that I remember, my mom just explained, you're just going to give extra blood and she said you don't have to but this can help someone else who's living with your disease in the future but you don't have to do this. There's going to be other kids who can do it actually, but you can make a difference if that's something that you want to do. And of course, then I was just like, okay! Sure, I'm going to get an extra sticker. That's all I really cared about, no extra sticks and I get a sticker. Looking back, that really just shaped the way I view research in that it can be an amazing thing and you can help other people, but just thinking about yourself at the same time and doing what's best for you, I think [is important]. She just phrased that to me in such a great way. I think with kids, it can be difficult because they are children and they might not be able to make an informed decision for themselves. But at the same time, it is their life and it is their condition. So I think each parent is going to handle it differently, but I think just being honest with your child and just explaining as much as you can that they're going to understand and when they get older I think explaining to them what research did they participate in and telling them that I think is also really powerful and letting them know what they did do when they were younger. I have so much respect for parents of rare disease warriors because they didn't have them and when we're young and we're not able to help and advocate for us in such an amazing way. I think just do what's best for you and as soon as you make a decision stick with it, and don't question it because you know what's best for your child and when they get older you could talk to them about it and it can be a really amazing and empowering learning experience for yourself and your child.

Imogen: Like I was saying, I watched the video of your speech at rare disease Day in 2017, and you mentioned that you're in college now and embarking on life as an adult.

I'm interested in how you deal with the uncertainty of having a rare disease now that you're an adult.

Anna: I actually graduated college in December of last year.

Imogen: Congratulations.

Anna:Thank you. Thanks. It is terrifying, but it's awesome. So honestly, just depends on the week, to be completely candid. Things changed for me a lot growing up. I think as you get older you learn how to handle things differently and what's going to work for you. For a long time, I dealt with the uncertainty of having the disease by ignoring it and just not acknowledging it and acted like it wasn't there. The more I got involved in things and made friends and friends started to ask questions, I realized that I couldn't do that. That wasn't fair to anyone and it wasn't fair to me because it was really me just being ashamed of what I was dealing with and there's no need to be ashamed of it. So now as an adult, it's a balance. It's definitely a balance. It's about being honest with those around you. My family, my siblings are such amazing support for me and it's been really helpful being able to go to them and talk to them about things and really hard conversations, you know. Like my sister has two amazing children. I love my niece and nephew and I never thought that would be a really hard time for me but it really was. I remember when she was having them, I was so scared they were going to have ALGS even though they had nothing to do with me whatsoever. Should I declare??? or otherwise, so I think just knowing that you're not going to know what's going to maybe set you off. You're not going to know what's going to come up and be a new barrier or a speed bump in the road. I think now I give myself a lot of grace. For a long time, I thought I had to be strong all the time and that's not fair either. I think I just deal with it day by day and just be honest with myself and those around me on how I'm dealing with things and when things are hard and tell people who don't have a rare disease what I do need because it's not fair to just be upset that they don't know [or] they don't understand; it's not their fault. I think

just being honest and open with others as well as myself is really how I deal with it at this point.

Imogen: That's awesome. And that's so great that you've had that support from your family. And that was going to be my next question. How have your family and friends supported you during you know hard times in this process and I was wondering if you could talk a little bit more about that and also whether they at times ever needed support themselves.

Anna: Yeah, of course. Yeah, my family has been my rock and that's still an understatement. I wouldn't be the empowered woman that I am today who happens to have Alagille syndrome without them. I joke that the biggest favor of my siblings ever did for me was to remind me constantly that I'm really just an annoying younger sister and I'm not ALGS. That's probably been the most powerful thing for me. I never knew I was different. My siblings didn't know I was different which is also just really funny. Looking back, when we were kids and people would call me short and they were like "what? No, it's just Anna. Like, what are you talking about?" I think that sense of security was really what they provided and that constant love and support. As I've gotten older I think It's grown even more so because now I feel like I have the words to explain things to them that they can't understand. I have the words now to express what I'm feeling when the uncertainties do get the best of me and I'm feeling really down. I can actually express that in a way that they can understand more so and they've been absolutely amazing for me and they're very honest. My sisters are really good and so are my brothers, just being like, "you know, I don't know what that's like, but I love you and that sucks and I'm here if you need to cry or yell or you know, whatever you might need." But I do think yeah, they also needed support. You know, a rare disease doesn't just affect the warrior, the patient, it affects the whole family. I think for a long time, it was really my sister that was the closest in age to me. She's only 14 months older. And so she kind of turned into my protector at school and she didn't have to do that and she… that was hard on her at times. Of course, she would never say that but it was hard on her

at times having to serve that role. Now that they're older and they're getting married and they're having children, we're having to have those discussions about this genetic disease and what that means and while that can be some really heartbreaking conversations, they know that they can come to me. They know that I will answer honestly. They know that my parents will answer honestly and tell them things and so I think it's really just supporting each other and being open and honest about things that are difficult. It's really funny, I think, sometimes I go to my sister about something and she's very caught off guard because there's still certain aspects about having about having a rare disease that I never used to express and now I'm starting to and sometimes she's just like what?? I think just being open and honest and I think a sense of humor always helps when it comes to family and friends and support during difficult times.

Imogen: That's so wonderful how supportive they've been and that's so nice how you can just be completely honest and open about everything. It sounds like you have had the opportunity to interact with other Alagille syndrome warriors. I'm interested in how you are able to connect with those people and how that those interactions have been.

Anna: There is an absolutely amazing nonprofit organization called the Alagille Syndrome Alliance. They are the one and only support system for those affected by Alagille Syndrome. When I was diagnosed, the founder, Cindy Luxhoj, her daughter Elena was only a couple of years older than me. I remember she connected my mom via phone and that's how Cindy got everyone connected was just calling people. Now it's an international nonprofit and we're connected to people around the world, which is just amazing. That's how my parents really got to interact. The first time that I ever interacted with another patient was when I was going into middle school. The Alagille Syndrome Alliance had an international symposium for those with Alagille Syndrome in Atlanta, Georgia. My mom took me because I was having a really hard time as I mentioned earlier so she took me in hopes that it would really help, and it did. It was really funny; I had never seen anyone itchy scratch their feet on carpets and stuff the way I did. It was just… I can't put into words how amazing that was for me, just seeing other people doing the

same things I did and not batting?? when I did my little dance on the carpet scratching my feet. But then when I got older, I connected with Elena Hans, Cindy Luxhoj's daughter, and a couple of other young adults and that was really when things really changed. I had kind of a pivotal point in my journey with the Alagille Syndrome right before I went off to college because in high school, I really talked about my disease. I only told those who needed to know and I was trying to decide how I was going to go about college and the place. I grew up in a small town so everyone knew I had something and I was going to a place where nobody knew anything. That was both empowering and terrifying. I was really trying to decide how I wanted to handle that and so I went to another symposium put on by the Alagille Syndrome Alliance. It was my first time going as a young adult and that was life-changing. I felt so empowered and I felt so… I only want to say normal. I just… I don't know it felt like coming home, connecting with those young adults and Elena and you know understanding that the struggles I was experiencing wasn't terrible, wasn't awful, and wasn't abnormal and that others are dealing with it as well. I do think the Alagille Syndrome Alliance is the greatest resource available to patients affected by Alagille syndrome. There are numerous Facebook groups. I actually started the first young adult with Alagille Syndrome Facebook group when I was in college in honor and memory of Elena after she passed away. There's groups for parents, there's groups for everyone but then their website is just a wealth of knowledge. We have everything available for parents who are going through school and their kids are going through school. We have classroom guides on how to navigate that because that's a beast itself without a rare disease. All of these things are available and it's… I can't even brag enough on the Alagille syndrome Alliance. They're really the reason that I am the advocate that I am today.

Imogen: That's amazing that you have such a supportive group like that. And it's wonderful that you were able to connect with those people and they helped you feel normal and I'm very sorry to hear about your friend. That's very tragic.

Imogen: She's no longer having to fight so it's okay.

Imogen: Taking all of what you've said into consideration, how important do you think it is for any rare disease warrior and their and their family to have a community around them who can you know sympathize and who's going through the same thing?

Anna: It's vital. I really can't say enough about how important that is. It's life-changing. I think the best way of putting it is, I did a fundraising campaign and was posting on Instagram a lot a couple years ago. One of my friends shared it and her profile is public. And so this this mom in the Spanish-speaking country saw it and saw Alagille Syndrome in one of the hashtags and had never connected with anybody ever. She started messaging me and I, of course, added her to Facebook groups and I told her about the Alagille Syndrome Alliance and she was sobbing because she was a single mother and her infant had just been diagnosed. He was going to have to have a transplant as soon as possible and it was just in the fight for his life and she was in all of that by herself. She went from fighting this terrifying, unknown battle to having an entire community around the world to support her in the matter of minutes. We're still connected to this day and it's just… that's what it can be. It can be the source of hope during a really really dark and terrifying time.

Imogen: That's an amazing story. That's so wonderful that you were able to connect with that woman and help her feel like she's not alone in all this. You've mentioned a few things that you've that you know, you started some Facebook groups and things which is awesome. I wonder if you could tell me a little bit more about your personal involvement in these advocacy groups and also maybe some suggestions of how other people can get involved

Anna: my involvement really started at that symposium I went to right before I went off to college. I was on the team panel Q&A at that symposium and actually the next year or the year after I was the keynote speaker at our symposium. That was really just how I started. I kind of just jumped in head first. I did start that Facebook group. It was right after Elena had passed away and I realized that a lot of us were grieving on different parts of the world and we were grieving separately. I was like, well, this is this is absolutely

ridiculous! Elene would be so disappointed in us, like “what in the world are you all doing? I connected all of you, I brought you all together and you’re going to grieve and go through this another obstacle by yourself. That’s just not okay.” That’s when I took it upon myself; I reached out to club members of the Alagille Syndrome Alliance at the time and asked for their help in finding people. It’s the first and only young adults Alagille Syndrome private group and so it’s just for young adults and that’s kind of where it took off. I just got bit by the advocacy bug and I haven’t stopped since. I joined the Alagille Syndrome Alliance as the first junior board member about a year after that, and was a full board member a year later. I was a full board member for about two and a half years. I recently had to resign because I have a job now, a full-time job, and so I am having to really focus on those things that I can do while working a full-time job that’s not patient advocacy. Thanks to people like Seth Rotberg who you also had on your podcast, I’ve connected with him and have attended a couple conferences. Really just meeting people, that’s really how it happened and just speaking my truth with my condition and supporting others with other rare diseases is really just how I got involved. I’m still learning. I’m still learning how to continue being involved. Now that I’m an adult with a full-time job that’s been a bit of an obstacle, but I’m overcoming it. I’m working through it. But I do think you know how you can get involved I think if you have a rare disease yourself, I think really finding that patient advocacy groups that you have and starting there, and starting with those who know you and know what you’re going through. If you don’t have a patient advocacy group because I know that that some rare diseases do not, if you’re a young adult, Oh my gosh! Everyone’s on Twitter. All the rare disease advocates are on Twitter. That’s how I keep up with a lot of them and I think just kind of keep on start??? and follow them, and retweet them, and tweet at them, and do things that way. I think advocacy looks really different for everyone so just find what works for you. For me, It’s telling my story because I’m in a place with my condition where I can do advocacy. I’m not fighting for my life right now. And so I’m going to do that kind of advocacy as long as I can on behalf of those with my condition who cannot. I think just find what empowers you and strengthens you. There are some amazing patients

that do art that is incredible to look at and just beautiful and gorgeous. Just know you don't have to do patient advocacy the same way that someone else does. Just find what works for you. Any type, level of advocacy is amazing and impactful. I think just getting started is the hard part but once you get started, it's hard to stop.

Imogen: That's great advice. Thank you and that's wonderful that you've been able to be so involved with the Alagille Syndrome Alliance and you know being on the board and everything, that's so cool. Also, congratulations on your job. That's very exciting.

Anna: Thank you!

Imogen: What is your job?

Anna: I work at a regional rape crisis and child advocacy center. I'm a prevention educator so I go and I'm trying to work out there and pulled out of a job????.

Imogen: Wow! So you're still doing some kind of advocacy.

Anna: Yes.

Imogen: You know, even though it's not rare disease related. That's really awesome.

Anna: Thank you.

Imogen: Congrats. I was wondering sort of overall, what is your advice for other rare disease warriors out there and their families who are coping with the rare disease. Where can they find hope?

Anna: Oh, What a question! I think a lot of times rare disease families are very hard on themselves. I think it's either you feel like you have to be hopeful all the time or you have to be positive all the time or you're stuck in a rut and it's really hard to find any hope. I think for those that feel like they have to be hopeful all the time, that's not true. It's not authentic and it's not real. I think just knowing that expressing the difficulties even to those who don't understand is half the battle and not being ashamed. I think

finding those key people who can listen and take the venting that you're doing and taking all of that and not give pity but give love and compassion I think is really powerful. I think for those who are stuck in a rut, I think it's just to remember that tomorrow's a new day. I think that is what really changed things for me is just realizing for myself. I'm a young adult with Alagille Syndrome and there's more of us now than there's ever been. However, there's not a ton of us and so I think that can be a really scary thing to look at and to know, to watch other young adults pass away. It could very easily just be depressing and just really suck you into that negativity and only focusing on that. I think just remember that you can't control the unknown. I'm a control freak so it really bothers me, but you can't control the unknown. You can control how you handle the unknown. You can either wallow at all these things you can't control and all these things that are happening to you or you can focus on the things that you can control, the things that you can change. I think just cut yourself a break; days are going to be hard and you don't always have to be happy but I think just you know, tomorrow is a new day and there's a huge support system out there just for rare diseases as a whole. If you can access it, I think that's where a lot of hope lies: the lot of amazing people around the world.

Imogen: That's really great advice. Yeah, I think it must be hard for people to find that balance between feeling like they need to be hopeful and finding those things that they can control. That's really thoughtful advice. Thank you so much. Well, those are all of my questions now. Is there anything else that you wanted to add or mention?

Anna: No, I just I can't thank you enough for having me on. t's been such an honor. I love sharing my story and having that opportunity so thank you so much for providing that opportunity. I really I really appreciate it.

Imogen: Thank you so much for joining me and sharing your story with all the rare disease community and all the best in your job. Hope it continues to go well. It sounds awesome.

Anna: Thank you. I appreciate it. You have a wonderful day.

Thank you for listening to this Rare Genomics RareShare podcast. Visit www.raregenomics.org and www.rareshare.org to find out more and join the community.