

## Introducing 1 in 20000

**Priya:** Hi! This is Priya, the editor of Suno India, a podcast platform for issues that matter.

A few months back when we started Suno India, a dear friend of ours Avantika reached out with an idea. She wanted to host a podcast series on rare diseases. I was excited and intrigued to know more and then, realised how less the conversation in the mainstream is around this topic. Most of you are wondering what is a rare disease and how many suffer from it. An estimated 100 millions Indians are said to suffer from any of the over 6000 rare diseases found in the world. So how is it that the conversation is so limited and restricted to only those who get impacted? This podcast 1 in 20,000 will answer this and many more questions. On World Rare Disease Day, we are honoured to launch India's first podcast on rare diseases.

**Avantika:** Hi! I am Avantika, the host of 1 in 20000, a show which will take you into the lives of those with rare diseases and give you an insight into the spirited world of those fighting to find solutions for them- from patients to parents; to doctors to geneticists. This show is brought to you by Suno India, a podcast platform for issues that matter.

**Priya:** So do you want to tell us a bit about what the podcast is going to be about, how you came about the idea of doing this podcast.

**Avantika:** I am super excited simply because this is the first podcast series that I will be hosting and this series is very dear to me. It's kind of the reality that I have been going through and it's so great that I have the opportunity now to tell people about this amazing cause that I'm working for.

**Avantika:** So the podcast will be on rare diseases. Now I know a lot of listeners would not know about rare diseases and that's the entire point of doing this podcast. Rare diseases is one of those things that we don't encounter in our everyday lives. And I am a person who has a rare disease, who has a rare condition. And the podcast series will actually be a journey for hopefully you as well and the listeners on what exactly are rare diseases and who are the people who are working with patients on rare diseases. So I hope it's going to be a blast for them as well.

**Priya:** I mean, the minute like, someone hears 'rare diseases' like, their mind will probably go in like, all directions. But like, are rare diseases usually hereditary? You know, what, I mean, do people know the causes, I mean, the cause behind it and how does a disease get classified as rare?

**Avantika:** Yes, that's a very interesting question, Priya, and to be honest, I did not know about rare diseases a decade ago. So, the reason why there's a separate classification of diseases as 'rare' is because the prevalence of these diseases is in very few people. So, if I just talk factually, the World Health Organisation has classified rare diseases as something which happens to one in a thousand people. So, that's the prevalence. So if the prevalence of one disease is one in thousand people that will be classified as a rare disease. So it happens in very few people medically, in very few people of a population, but collectively, the numbers just go phenomenally high. When, if you consider WHO UN statistics themselves, there are 6000

plus rare diseases in the world, and this is just an estimate. Another estimate says eight thousand. when I got to know about this, I was just mind-blown. here is a mass of people about millions and millions of people if you put all the rare diseases together and nobody is talking about them in India. Well, at least, let me be fair, it's not like nobody is. But the conversation is very low. And very slow.

**Priya:** Yeah, I mean for an estimated hundred million people with rare diseases, I think the conversations are not happening even, you know. So, just a bit of backtracking. When, and you don't have to go too much to detail, but could you perhaps tell us when you found out that you have this rare disease?

**Avantika:** Yes. Yes, yes, of course, why not. So, I was a very happy, healthy, chirpy-- and I still am-- you know me for years now. had no symptoms whatsoever. And in my...so my childhood was spent relatively well, with very minor sort of health issues. But in my teenage years, I got...one fine day, I was in the doctor's office because my physical health deteriorated a bit, and we went to the doctor,, and the doctor saw me and said, "I think you have this rare condition." And I was flabbergasted! Because I was a teenager! think hink 16? And barely like, out of school. And I did not know what rare diseases were. So that was my first brush with a serious chronic medical condition. My medical condition, from what I know, and I will come to the definition of it later on, and how people know they have rare conditions or how medical practitioners diagnose their conditions later on perhaps, but...as per one estimate, only one in twenty thousand people have what I have. And, most of them are boys. The thing with rare diseases is that rare diseases happen at any point in a person's lifetime. So it...for me, it was, the onset was teenage years. But eighty per cent of the cases are genetic and half of these...so half of the rare disease sort of pool occurs in childhood. So, imagine if you're a child and going through something which is way beyond your capacity--emotional, physical to cope. That's just....really, really intense.

**Priya:** So yeah, I know we will be going a bit, you know, more into this conversation a bit later. So could you perhaps tell us, what it is that listeners can expect in, from the series itself. You know, who are the kind of people who will be part of the series and yeah, and what do you by the end of it hope to have achieved?

**Avantika:** Yes, so I don't think there are easy solutions when it comes to rare diseases. The entire intent behind this podcast series and the number of episodes that we will be bringing out is to start dialogues on this very critical issue. As I said, not many people know what rare diseases are and imagine if there is somebody suffering from a rare disease around you, you would never come to know that they have that. In many cases, patients themselves don't know they have that because the level of awareness is so low. So this is what we're talking about. This is just this very small step that I and Suno India and through you, I'm taking. So that people just have a primer and an introduction on what rare diseases are. But more than that, it's also for insightful conversations on these extraordinary men and women who are working very day on rare diseases. I mean, we don't come to know about them, do we? So, there's genetic work being done on rare diseases. Unfortunately, not a lot is being done in India but abroad, it is scaling up. And in India, there are social rights activists, lawyers, working specifically on rare diseases and disabilities and discrimination. So one podcast will be entirely on the legal rights of people with rare diseases or in general, people with disabilities as well.

Then, another podcast will be of this extraordinary person who has built his life with a rare disease. He was perfectly normal, again, and in the course....now he's middle aged, but in the course of his life, he progressively deteriorated and unfortunately or fortunately, as he puts it, "this is what it is and it's not a bad thing as such." And so he is now bound to a wheelchair but the amount of life this person has is extraordinary. And has a family history. Like, I did not have a family history. But he has a family history. So two of his siblings also suffer from the same rare condition. And they are now coming up with a hospital in Solan for rare diseases. Specially muscular dystrophy. So one podcast will be on that as well. We will be talking the medical condition of rare diseases -- what the medical sort-of jargon says, how geneticists work, and what is being done in that sphere.

**Priya:** Avantika, could you tell us a bit more in terms of what is the status, or what is the situation around research, policy and also understanding of rare diseases in India itself? I understand you said there are anyway between six thousand to eight thousand rare diseases in the world. Do we have, like, a register or something where, you know, the identified rare diseases that 'in India, there...' Like, what's the status in that sense?

**Avantika:** I don't think the government knows itself. So there has been a rights movement--disability activism as well. And rare diseases as well, rights movement, for the past couple of years. Which is how huge rare diseases have now been included in the national disability policy. The government came up with the national policy for treatment of rare diseases two years ago, that is a good step. Last year, they, if I'm correct, last year, they issued a gazetted circular, which meant that the policy is going to be revised. And that's a very good step in my view. Because they're trying to keep pace with medical innovation, they are trying to keep pace with the needs of the society as well and the needs of the patients. I'd like to give a bit of background on rare diseases itself before sort-of focussing on what India is doing. So as I mentioned earlier, rare diseases can occur any time in a person's life....without any symptoms or family history. What really happens then is that the patient won't know what's happening, the family is unlikely to know what's happening so who would? Medical practitioners, doctors would, ideally. But that's not always the case. So people who live in the cities have more access to hospitals and super-specialised hospitals, if that. And if you consider in a rural area, it is highly likely that either they will be misdiagnosed or referred to somebody who is not specialised or who does not have the adequate medical knowledge to diagnose that rare disease. So misdiagnosis of patients is very common in this field. Also, the number of patients that India has is something that is best an estimate. India does not have a solid definition of what rare diseases are. We have not settled on that definition, which means that according to some estimates, seventy two to ninety six million people in India are people with a rare condition. For that many people not be having a cohesive, coherent policy is very disheartening. Secondly, not be having the right kind of medical infrastructure simply because rare diseases...the nature of these conditions are genetic, which means there is no cure. The maintenance of the quality of life of a patient needs a lot of financial investment. Sometimes, it can go into lakhs of rupees. Which somebody who is of a certain economic class, whether it be low, middle or upper middle--maintaining that kind of treatment is out of the question. So they get sidelined. Without the family....what can the family do? So...all of these are really big questions that stare us in the face right now.

**Priya:** I think having like, a national or a state policy around rare diseases will also ensure that

there is you know, the whole inclusion aspect of looked at, right? And covered. Because like you rightly pointed out, like, there are very few of us who even know of people who have any sort of rare diseases. And it could be that because either they are not identified or because society is so structured that there is some amount of invisibilizing of these people happening, if you get what I mean. Like, you know, like, lot of times--and this is something that I have, I used to cover a lot of you know, when I was a reporter for The Hindu, I used to write a lot about disabled people and...one of the things that I realised as a course of writing about it was how the structures-- infrastructure, or even the way things are just structured as a society doesn't really give that kind of visibility for them, right? I just feel like you don't, like how many you know, how many visually challenged people do you see walking on a day-to-day basis outside? Or, people with certain kind of disabilities or people on wheelchairs? Like, there is certain amount of, and you wonder like, where are all these millions of people? Why don't you see them in malls? Why don't you see them in museums? And then, you know, if you reflect, you realise that the need to be "normal"-- you know, the need to be "normal" is just celebrated so much that anything off that normalcy has absolutely no space.

**Avantika:** I completely agree with you I think that was my struggle growing out of that...so after I was diagnosed, it took me five to six years to get out of that entire trap of whether I am abled, differently abled. Whether I'm a person with disability...what am I? And also, it has a huge impact-- the way the society around you is structured has a huge impact on the family and on the person going through this as well. Because not only are you dealing with something which is way beyond your capacity, but each piece of your life-- whether it is work, or whether it is education, or whether it is the government policy, or anything else...personal relationships. It's just catered to able-bodied people. And I don't know how fair or unfair that is, what we are losing out on is this mass of people who have such incredible talent, and who can contribute and participate in the society on an equal footing. But just because we, firstly, don't know about, don't know enough about-- which is on us, mostly, actually-- don't know enough about rare conditions. Secondly, we may also be very, in our heads, very rigid. On what the normalcy looks like. And, in that sense, the loss is everybody's. Not just people with disabilities. And not just people who do have rare conditions.

**Priya:** But before we wrap up this conversation, a quick question: why did you decide to do this podcast at this point in your life?

**Avantika:** I had been thinking of doing a project for the longest time. And an obvious reason is that I got in touch with you and everything panned out. But a more in-detail answer would be that discussion on rare diseases need to be had now. The children-- we've already covered the children aspect, right? That children are adversely affected by rare diseases as well. These children are going to grow up someday. We need to decide as a society, are we going to lock them up in homes and not let them be? For instance, when I was growing up the terminology that was used was 'handicapped.' Now I'm not talking just of rare diseases because usually rare diseases they do lead to one or the other kind of disability. But, now, you tell me Priya, do you think you would want to call your friend as handicapped? I know I wouldn't. And...yeah. And, then people sort-of thought about the world itself and the language itself, and how people with disabilities were seen. And they came to the conclusion that handicapped was a word that people used for other people but it did not accurately reflect the circumstances of a person with disability. It did not accurately reflect what the person's

life is like, what their needs are. And if we are speaking of millions of millions of people, that's a huge loophole that you're looking at them. As a society. And, in due course, the language changed. If I just take that instance. From handicapped, now it is persons with disability. Not disabled people, or anything else. 'People with disability' is the officially, most accepted usage simply because people who are perceived as disabled, might not actually be disabled. Physical disability does not mean emotional or disability at that. So, there are so many grey lines-- sorry, so many grey zones in that rare diseases throw up. It is very necessary, I feel, that we start having these conversations now.

**Priya:** I think like, we were having this conversation about while it is good to have a classification of rare diseases and what this means, I think the nuance matters. Like, we have been talking about it, I think, in private conversations while we were talking about the series, I think. Who are we making these classifications for and what is the purpose of it? Is the classification so that there is easier and better identification of disease itself thereby benefiting the patient or is it more, you know, more for the government policy itself? I've always struggled with that question. we adopted our daughter and I remember when we had to register-- when we were registering and sort-of choosing the age-group, there is a column where you can tick for children with special needs. And we didn't choose that. And because we wanted a normal child, you know, a normal in double quotes. Because there was also this, because we didn't know what special needs would entail. What kind of, financially, what would that mean for us? And even in terms of treatment. And this is us, like, fairly urban, you know, privileged young Indians. And having fairly okay access to information. I was telling you, most of the children who get adopted with special needs, they end up within the inter-country adoption pool. And I think as of last year only, forty seven children with special needs were adopted within India. Whereas I think over three hundred and fifty children plus were adopted outside India by you know, foreigners. And then so there's this huge push now within the adoption community to have a classification in terms of, you know, classifying the needs in itself as, mild to moderate to severe needs. And I'm always like, thinking at the top of my head like, but who are they classifying this? Is it for the parents or are they doing this for the children's sake? So I just, you know, this is such an important conversation but also a difficult one to have.

**Avantika:** Exactly. And I completely see your point here. Because these are the exact same conversations, for instance, when you referred to the point of classification. These are the exact same conversation that the government is trying to have with activists as well. And there are no easy answers. There are no easy answers simply because rare diseases is one of those issues that humankind faces that tie in so many areas. Morality, ethics, society, interpersonal relationships, legality. So, if you ask me, these classifications are quite a lot for procedural and medical purposes. But they are not exactly patient-centric when it comes to society in itself and how patients and people with rare diseases or disabilities function within the society. A few decades ago, when people used to be prescribed spectacles, that was considered a disability. So that meant that if you were a woman, your marriage prospects just decreases by a huge margin. And I was just like, what? [laughs] Because now, you see people wearing spectacles all the time every time, and every other person that you see is wearing spectacles. So the entire notion of disability that existed then isn't existing right now. And which is why I think that somewhere we are also late in having conversations on rare diseases as a society simply because not just the number of people that might be suffering from or

might be diagnosed with a rare disease as we speak has increased, but on the ground, what do we have? We have possibly policies, yes, the government is trying to do what it can. It could do more, that's what activists say. And I'm not even getting into that debate. But at least some semblance of a policy is there. However, is it a coherent policy? And, is it being implemented on the ground? And for that, I would for certain say that no. How many of us who have rare conditions know of our rights? I didn't know. However, as I mentioned previously as well, the people who are trying to bring a shift in thinking of patients, of families, of medical care providers as well as those who are engaging with the government-- they are phenomenal people in their own right. And I hope to bring those stories out and those instances out, so that after the podcast series, in the long run, your listeners could possibly see a child and say that, hey, maybe, this is a child that might need some more help than in quotes, normal children. But if we provide that opportunity, this child has the future to look to where he or she can shine. And that child will not be going through the same sort of rigours that patients with rare diseases mostly go through. That what has happened to me? Where can I go? Where can I seek medical attention? and something as basic as Will I survive this or not?

## Conclusion

**Avantika:** Thank you for listening to this episode of 1 in 20,000! We will be back with another episode soon! You can listen to this podcast on Itunes, google podcasts or cast box. Don't forget to check out [www.sunoindia.in](http://www.sunoindia.in) for podcasts on other topics! As independent producers, we rely on you to spread the word! So show us some love by sharing this with your friends and family! Don't forget to use #1in20000 on social media while sharing this episode.

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