Episode 2 - Care for a rare
(1 in 20000)

Hello and welcome to the second episode of the 1 in 20000 series. First of all, thank you for the amazing response to our first episode, India. Please do keep sending in your feedback. Today, I am going to introduce you to Mr. Prasanna Shirol, the founder of ORDI, a rare disease patient care and advocacy organization. He is an Ashoka fellow and has extensive practical experience having been associated with the cause over the last 20 years. In this podcast, we will talk about what is being done for patients with rare diseases, the status of rare disease patients in India, and his work to create a better future for them.

Avantika - Welcome to the show, Mr. Shirol, we are glad to have you onboard. I have a lot of questions from you and hopefully we will have a very informative session in the next 20-30 minutes.
So my first question would be, why did you start ORDI and can you tell us a bit more about your personal experience?

Prasanna - ORDI was started in 2014 because there was no common voice on rare diseases in India representing all the 7,000 plus diseases, and the challenges of these patients, families and caregivers. There was a need to unite patients, smaller disease support groups and to bring all the stakeholders on the common page, to arrive at a common voice on the requirement of the community etc. So, to address these challenges, ORDI came into picture. but However there is a long history personally, my journey started with my daughter who was diagnosed with Pompe at the age of 7 years after going across India visiting various hospitals and 40 + hospitals and doctors, we diagnosed it as Pompe when she was 7 years. Pompe is a neuromuscular disease where due to deficiency of a particular enzyme leads to muscle hardening and tightening. So, when she was born she was normal. Of course there were initially we had problems of infections- lung infections where repeated hospitalizations and visits to doctors. However, the doctor felt that it is common in some of the children, and it may take some time. But our worries increased when even after one year she was not normal in terms of her activities - getting up, crawling., Then at the age of one and a half years or close 2 years we went to NIMHANS and diagnosed it as a Myopathy or a GSD Glycogen Storage Disorder type 2, it was only a suspicion but not confirmed diagnosis. At that point of time, doctor told us this is progressive disease and your daughter may not survive or she may not be normal after a certain age and she will get bed ridden. So, with this now our journey started, and we finally got it diagnosed as Pompe. She started respirating on an artificial ventilation through tracheostomy, and then when we started looking for other options by that time it was taxing on the family burden then the professional career then financial burden and various things but however we continued the journey and through international Pompe association we were able to get in touch with a company who offered to give enzyme replacement therapy, the only approved therapy at that point of time, of course even today and we got access to it. After the treatment, there was a slight improvement and of course she continued to be on ventilator but she was normal in terms of our metabolic activities and some improvements were there. Then next challenge was to make her inclusive because mentally she was fine so we started going back to school where my wife used to go along with her, stay back in the school whole day and then bring her back so the journey continued and in between we also had major interventions like due to the long term medications she had to undergo therapy costing in lakhs and we overcame about that then because of the neuromuscular sensitivity she also underwent scoliosis surgery, which is one of the longest and toughest surgery. So these are some of the challenges in terms of management, supportive care which is taxing both mentally and financially because there are no any healthcare policies or insurances which are covered under this. So like this we pulled on and then she started going to college also and on wheelchair with ventilator she was studying in Bcom 1st year which is the Degree 1st year after 12th and unfortunately the ventilator conked off when she was in college first year, and she had a

cardiac arrest. Then last 16 months now she is in of course she has come out of coma but he is in a semi coma stage, and we are at home and she is at home and my wife is taking care of her. So the entire journey made me to think: 1) how do we manage such children with various challenges because everybody in the family gets affected and our career and dreams get shattered? Then what is the long term solution is one question? Second question, if the company is giving free medicines to my daughter what happens if they stop? For how long they will continue? And the third question was when I got this support from International Pompe Association, then I understood the power of parent support group so there was no such system available at that point of time in India and I thought we should do something.

Avantika - Indeed! In this conversations with you, you spoke of building a movement now I am curious what is the movement about and why is it needed?

Prasanna - See if you look at any big change in the society approach, and if you have to get the government support especially in a country like India with various challenges and if you have to get their attention we need a big movement. Similarly, for rare diseases also if you look at the past experiences of other landmark societal changes including the recent one of Article 377, it all started with a big movement. So similarly a movement was required to sensitize on the condition, requirement and challenges, and status ofrare disease community was needed. Then our requirements are different, they are long term in nature, they are multidimensional and expensive, and they are taxing on the entire family not just the patient. These are not addressed by any existing healthcare policies of Indian government both state and central. there is a stigma associated with rare diseases and need to overcome these then we need to consider rare diseases as a medical challenges under the PWD Act that is person with differently abled act or a separate Act focused more on medical and invisible challenges. So these are the few top notch requirement of our rare disease community, and definitely a movement is required because one it always says village require to make a change.

Avantika - Could you tell us a bit about the policy part ? In your experience what are the some of the challenges in convincing administrators that their attention is required on rare diseases?

Prasanna - More than challenges let me brief you about initially the work which has happened in India in this area. Initially when I started working, with more than 12 State governments in India so only the Delhi government responded to certain extent and recommended a financial support then the health ministry turned it down, the common discussion with the policy maker is biggest is the cost they say for example while we have some diseases or conditions requires around 2 lakhs or 4-5 lakhs per year in giving a proper diet to some set of diseases called IEMC (Inborn Error of Metabolism), then we have certain conditions which require one time interventions to the tune of 25-30 or 40 lakhs for the bone marrow transplantation or a lung transplantation etc but the other extreme end of rare diseases where the treatment is available most of these treatments are very very expensive and these treatment are given based on the body weight of the patient. For example, if the body weight is 10kg, the medicine required, the cost may be around 30-40 lakhs per year life long. So, similarly when the patient increases, grows and the body weight correspondingly cost of medicines per year goes upto 1 crore 1.5 crore and so forth. So, these are the challenges which the Health Ministry was not equipped to handle. Then second part is the numbers: they did not know or even today they don't know what is the burden? Where do they start with? And what happens if they start giving treatment to one child or two children, (how) the burden on the exchequer goes up. Third thing, the focus of the current or existing government is always focussed on addressing the primary healthcare challenges because still today we talk about general challenges of scarcity of doctors, scarcity of paediatricians, primary healthcare challenges there is no hospitals in the remote villages. So entire machinery is focussed on those and basically now focused on the number driven approach. For example, if there is a huge number of particular disease burden then everybody runs behind that and similarly the government.

But in our case, while we talk about 7,000 diseases individually they are very very small. For example, some diseases in India could be you know 1 patient, 10 patients or few 100 patients, that too, you know, spread across India. based on the PIL filed by the patients, Delhi High Court ordered a policy based on which the central government came up with a policy in 2017 but later in 2018 probably in December they kept the policy on abeyance because they understood the challenges of rare disease are still higher and they felt the policy needs to be reframed. So now they have taken 9 months time to reframe or redraft the policy which is the first setback to the patient community. Then later we also filed a PIL in the Supreme Court and we are now waiting for this. The big change what we have brought after filing the PIL in supreme court is that earlier there was a discussion between only the High Court and the Central government and few states like Karnataka who were interacting with us in terms of providing some solution. But since health is a state subject there is a need to involve all the state government and union territories into this discussion and our based on our PIL when the supreme court has sent notices to all the state government and union territories so that everybody takes the ownership

Avantika - you mentioned a term called "burden." Now, as I understand it, burden is public policy speak and medical jargon for the number of people with a specific condition is that right? So it is not actually you did actually touch upon the aspects of how it is difficult for patient and families to cope but is it right to think about patients in the sense that they are a burden?

Prasanna - See definitely they are not a burden because legally or personally nobody is a burden unfortunately this is the terminology used by the policymaker, you know health care burden or exchequer burden But legally if we speak everybody has the right to live and everybody has right to health so why should we deny the chance of living for a child or a person only because they are few in numbers and only because treatment is costly?

Avantika - That is a beautiful thought. I do agree with you, and that is one reason why we are bringing out 1 in 20,000. My next question would be on the patient advocacy part. You are in touch with patient advocacy groups abroad as well, and I was curious on what is the status of welfare of patients is there and what do these groups share about conditions in their own countries? How are patients faring in countries abroad, and here I am talking about a bit more advanced possibly or if you have experience of developing countries as well? That perspective will be really nice to have.

Prasanna - one of our major objective is also to work in creating disease specific patient support group in India because when there is no expertise available on medical fraternity in terms of management only when the families come together and share their best practices in managing their children and work with the medical fraternity we can come to a conclusion saying okay these are the set of protocols or list of things which need to be done or not to be done for managing the particular patient. So patient group holds an important role and our objective is to create one also and work with the existing. So far we have been working with more than 20 patient advocacy group in India, and also we are in touch with our work with Asia, Europe, UK and US. As you mentioned, the challenges in the other countries are slightly different. So let's look at the example of US. In 1983, when the Orphan Drug law came it was focused more on new drug development, research and development because in US there was no problem taking care of a child or healthcare facilities in terms of be it primary secondary or a tertiary care, and finally when certain things were agnosed only requirement was to treat them. But in India we always talk about rare disease policy and we always advocate for separate rare disease policy which is focused on patients requirement right from healthcare challenges, identification, screening then rehabilitation. But we also request and lobby for a separate orphan drug law in India because of the huge population. So this is one major differentiator between us and others. Then in other countries, yes, over a period of time, and probably they are all some of them are smaller

countries, there is an evolution where in some countries there is a rare disease policy, in some countries the policy is not there but certain conditions are considered for reimbursement and treatment is beginning. But there is a process which has started from long term and it is being reviewed every year and more and more patients and more and more rare diseases are being supported. But in India that is not the case, it is just the beginning. when you come to patient group if you look at you know European and US now most of the patient groups who have done extremely well in terms of creating an awareness and arriving at funding they all have started investing and or funding for research and development of a drug for their conditions because the general industry may not come and invest due to the... again the cost, huge cost involved in development of a drug, and also cost need to be recovered by very few patients worldwide. So obviously patient groups are taking initiative, and there are very successful example where patient group has already got an FDA approval drug for their diseases. But in India still we are fighting the basic primary healthcare need and challenges. Then if you talk about different conditions of their own country, the challenges are similarly the same, you know every patient goes through the same trauma, same problems but the only thing they address much faster than India. Then if you look at the smaller countries in Asia itself - Taiwan, which is the smallest country, has a rare disease policy as early in 2000, and very recently we have some initiative coming from China and Japan has almost you know full-fledged rare disease policy. and very recently in Malaysia and Singapore about also there is some movement from the government and I think Overall we are seeing a very hopeful situation across the globe and also in Asia

Avantika - That is very heartening to hear I must say, and associated with your last comment what can or how can people support your movement, ORDI and the cause of rare diseases. Could you let us know?

Prasanna - In fact this is one of the major area of either ORDI or any non-governmental organisation depends on. The support from various stakeholders is very, very important. On a long term basis, we have major programmes like for example we started rare disease helpline, so we need support. Then we have a rare disease care coordination centre which is a holistic management of patients, right from identifying, diagnosing, treatment and rehabilitation. Then we have started Home for Rare which is India's first assisted and palliative care for the rare disease patients and families. Then we have several advocacy meeting where there is need to be travelled across India. Then we have awareness program like Racefor7. definitely we require funding without which it is not possible so these are the many activities that require funding in absence of the Government support and we keep losing many patients childrens everyday so definitely we need support from the donors this is the financial part of it we need connectivity with the government policymakers be it the bureaucrats or the ministers through their contacts so that we can reach out faster, minimising the wastage of time. we need volunteers to talk about rare diseases in various forums so that it will help us to create a movement we also have a program special program called Pragati this is a family mentorship program where each person adopts a family and mentors and try to bridge their social gap because what happens when the family diagnosis the rare diseases they get into a shell saying ok we are the only people in the world who got affected and the cut off their social activities there are a lot of needs they don't speak to the outsiders so this person who mentors them bridges the gap between ORDI and the family so that he focuses them on personal basis and try to understand the needs and communicates with ORDI and maybe help them in small, small care so these are the programs both long term and short term we are financially and non financially we seek support from various people Finally, I can say that anyone who says I care for a rare is required for serving this community.

That is a brilliant thought to end this episode with. Thank you so much Mr Shirol. I think you are doing wonderful work. It was such a delight speaking with you.