

## **Interview: Dr. Sheela Namboothiri**

I am Dr. Sheela Namboothiri. I am working as a Clinical Professor and Head in the Department of Paediatric Genetics at Amrita Institute of Medical Sciences, Kochi.

### **Tell us something about the high incidences of genetic disorders in India.**

One of the main reasons is the positive consanguinity. There are a lot of people who marry blood relatives and it is very prevalent in certain parts of India, especially in southern parts of the Kerala and northern parts of the Kerala. Other places where the consanguinity is very prevalent is Karnataka, Tamil Nadu and Andhra Pradesh. Whenever people marry first cousins or even distinct relatives what happens is that the chance of them to share the defective genes go up. There is a high chance of them to have children with genetic disorders. This is one of the reasons.

### **Even if there is no treatment option for genetic disorders, do you think genetic diagnosis is relevant.**

It is very important because once you have a concrete diagnosis then only you can offer prenatal diagnosis for this couple. If they have one child who has a genetic disorder for which there is no treatment is available the parents are extremely upset. Another thing is that they go from post to pillar for their diagnosis and they have real guilt in their mind because once they go to many hospitals what happens is that they know that it has been branded as a genetic disorder. So they think that they have given a defective gene to their child. They want to know and we should communicate with them saying that we are all human beings and we all have defective genes in our body. It is not their fault and this takes off the guilt part from their mind and definitively it is extremely important for them to have a concrete idea so that at least they need not run around and they can do what is useful for that child.

### **How do you see the treatment options for genetic disorders?**

Even though people have a belief there is no treatment aspect which is available for genetic disorders, currently there are certain genetic disorders for this treatment options are, very popularly, available. One is lysosomal replacement therapy, enzyme replacement therapy, is available. Currently, we are treating 11 patients with lysosomal storage disorders in our own department and there are certain disorders something like cystenosis. Metabolic disorders definitely have treatment options if they have been diagnosed at the early stage and so it is extremely important to make a diagnosis at a very early stage so that the complications can be preventive.

### **Tell us something more about the department of Paediatric Genetics at Amrita.**

I have been working as a paediatrician for 12 years and then I got interested in genetics. I had my post graduate training in Genetics from the University of Glasgow from Scotland and came back and started this department in 2005. This department has been there for the past 11 years and I am extremely happy that now we are able to help families with genetic disorders mainly from the point of view of making diagnosis and also for prenatal diagnosis. This is our main agenda and now I am also happy that many patients are being referred from throughout Kerala, not from Kerala alone, and from other neighbouring states as well as from abroad. In Kerala, what happens is that people are very much health conscious and there are many many self referrals because now people are much more aware of the genetic disorders and the need for making diagnosis at an early stage.

### **Tell us more about skeletal dysplasia.**

My main area of interest is skeletal dysplasia. Here what happens is that this group of patients who have short stature, who are having curved bones and those who have difficulties mainly the short stature and curvature of bones. So this type of patients were always neglected and all were just clumped into one group of genetic disorders like skeletal dysplasia. But this

group of conditions by you have more than around 200–300 types of skeletal dysplasia. The main matter is that you want to know what exactly the patient is having and so that at least we can prevent it in the future pregnancy. The other thing is that there are some type of skeletal dysplasias for which there are some treatments which is available mainly in the form of a surgical repair and so that at least their morbidity can be reduced, so that they can have a better life.

### **Can you please tell us about lysosomal storage disorder?**

LSD is a condition where you have something like more than 50 disorders for which around 7 conditions there is treatment. In the earlier question also I have addressed this issue. The main treatment is in the form of enzyme replacement therapy and currently for 5 conditions we are treating patients with LSD in our our hospital. It is mainly in the form of their conditions are Gaucher's disease, pompe disease, and mucopolisaccharidosis type I, type II and type III. In these 5 conditions, what happens is that you want to make a diagnosis at an early stage because the permanent damage has happened to the systems then it is very difficult to revert the systems back. The caveat here is it is extremely costly so patients can't pay from their own pocket. In India the main problem is that patients are paying from their own pocket and this lysosomal storage disorder is the treatment option that is mainly it is being taken care of by the companies at present. So in western countries, what happens is the government is taking care of the responsibility but in India it is still in its infancy and it has not started.

### **How about the syndrome in your name?**

There is a syndrome is there it is called as Namboothiri syndrome. There are around more than 30,000 syndromes which have been named and when a new syndrome is identified which can't be fitted in with any of the norms what happens is that it should be publicized in an international journal and it should be presented in the American Journal of Human Genetics meeting were the experts will be accessing whether it is a completely unknown syndrome and then at some point in time that syndrome will be include into the London Dysmorphology database.

There were 2 patients who had come so many years back from northern parts of Kerala. They were siblings and both of them were having some abnormality of the feet and they were having severe mental handicap with changes in the face which were not described before and all the other conditions which could have some similarities with this conditions were all ruled out. It took some round around 5–6 years for us to formulate the condition and even now the gene has not yet been diagnosed. So we are in the process of identifying the gene which is responsible for this condition.

**Tell us something about Amrita's role in supporting children with Down Syndrome.**

We in the past 11 years have seen around 750 children with Down syndrome and it is extremely important for having to do something little extra for these patients rather than saying that you have the condition for which there is no cure. There are so many families that are so upset, so we wanted to go a little ahead and we wanted to have a support system or support group so that the parents can have a helping hand from the other families who all had gone through the same situation and how they cope with this situation. It is named 'Prathyasha,' which means which means 'something to look forward to' in Malayalam. So here we conduct meetings around twice a year where we have call the families and we call experts who are handling children with Down syndrome and actually we are having our next meeting on 26th of this April where experts on medical care will be speaking or those who are handling children with disabilities will be addressing them and at the end of the program we give an opportunity to the children to express their talents. Every year, I think the families really looking forward to that situation there as they find it very interesting to come and attend the function.

**The message that you want to give to the paediatricians and others who deal with multiple abnormalities.**

That's a very important question, which I wanted to address because in many situations where a child has some multiple abnormalities. What happens is that it always written as

multiple anomalies. In that way, it is not going to be advantageous at all once a family comes back to see a geneticist because we don't know what exactly the other child had. So if a child has been very severe abnormalities what definitely that child should be showed to a paediatrician and all the genetic features which you can see from outside should be documented and everybody has a smartphone so we should always take a photograph of the child and x-ray. These are two simple things which can be done by anybody. So things will help a lot because rather than saying that this child have an abnormality of the hand if you can see it in a photograph or if you can see what exactly was that in a x-ray. So this will be helping the geneticist in making a diagnosis in many situations and mainly in the skeletal dysplasia. It is always advisable whenever you don't have a diagnosis it is always advisable to store a sample of EDTA blood sample to be stored for future DNA studies. If at some point of time we need it will be very useful.