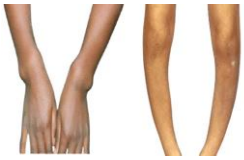
INFORMATION ABOUT ONGOING TUBULOPATHY REGISTRY

Study title: *Setting up of registry and targeted exome sequencing in children with renal tubular disorders.*

If your child is not growing.	
If your child is drinking a lot of water.	
If your child is passing a lot of urine.	
If your child's wrists are bending and bowing of legs.	
If your child is forming recurrent kidney stones.	

So this is a serious topic, if any of these symptoms are seen in your child, then your child may have a disease of **Renal Tubular Disorders**. It is a serious disease and if this disease is not treated on time, your child also remains in danger of life. Read the information sheet for information related to this disease and its treatment.

INFORMATION ABOUT ONGOING TUBULOPATHY REGISTRY

Study title: *Setting up of registry and targeted exome sequencing in children with renal tubular disorders.*

1. **Purpose and background:** Your child is having renal tubular disorder. Renal tubular disorders are a group of hereditary and acquired diseases that involve complex dysfunctions of transporters and channels in the renal tubular system. The disorders may lead to fluid loss and abnormalities in electrolyte and acid-base homeostasis. Normally, the kidneys remove excess acid from blood, but certain diseases, genetic defects, or drugs can damage a kidney's ability to do this important job. This can allow too much acid to build up in the blood and cause problems. When this happens, it's called renal tubular acidosis (RTA), the most common disease among tubular disorder**.** Without treatment, tubular disorder can affect a child's growth and cause kidney stones, fatigue, muscle weakness, and other symptoms. Over time, untreated metabolic abnormality can lead to long-term problems like bone disease and end-stage renal disease, requiring dialysis and transplantation. We propose to conduct studies on the inherited defects in the pathogenesis of tubular disorder that evaluate the entire coding region of patients' genetic material in order to identify defects that may be responsible for causing tubular disorder and to track medium term outcomes (renal dysfunction, growth retardation, nephrocalcinosis, hearing loss) of children enrolled in registry. This study is expected to provide much needed answers to unaddressed questions about disease biology.

   Therefore, we are creating a web based registry where experts can share their experience on the course and outcomes of therapy for pediatric patients with tubular disorder. The participating partners can collate their responses and reach consensus on strategies for clinical practice. Most importantly, however, we shall maintain a patient registry in which information about the disease in children with tubular disorder shall be collected regularly. This shall allow the partners to understand the underlying conditions, recognise the pointers of severe disease, and to compare how their patients are faring on certain therapies compared to other patients treated in the network centres, some of whom may be being managed quite differently. **The standard treatment of tubular disorder shall be instituted as in other children diagnosed with the condition.**

2. **Procedure:** Your child is invited to take part in the research study involving a group of children with *renal tubular disorders* in India. With your approval, your child shall undergo blood and urine investigations as required routinely for diagnosing the condition. This shall include testing for blood levels of hemoglobin, urea, creatinine, albumin and cholesterol, estimation of protein and creatinine content of the first morning urine and blood gas analysis. For purpose of the study, one additional blood test shall be necessary, for which about 5 ml (approximately one teaspoonful) of peripheral blood shall be drawn. Parents and extended family members of children, diagnosed clinically with the disease, will also be included.

Except for the latter additional test; **all other investigations are undertaken routinely in the evaluation of any child suffering from tubular disorder**. **These evaluations shall be provided free of cost**. The details of disease and treatment will be recorded.

3. **Potential side effects/complications:** Peripheral blood sample (5 ml) collection will cause mild pain locally in the arm and will pose no harm to you or your child.

4. **Cost and compensation:** You will not receive any monetary incentive to take part in this study. However, the blood tests and being performed for this study will be done at no cost to you.

5. **Confidentiality:** All the medical information of you/your children will be kept confidential and only authorized person who are involved in the care of your children and those involved in this study will have access to it.

6. **Potential Benefits**: This study will provide better understanding of the pathophysiology of the illness, predict disease course and outcomes, and improve care of your kid/relative as well offer antenatal diagnosis and genetic counseling where appropriate.

7. **Contact person:**
   In case of any query regarding study, you can contact us at nearest centre.

   - Dr. Girish Chandra Bhatt, Department of Pediatrics, AIIMS Bhopal, M.P.
     E-mail: drgcbhatt@gmail.com
   - Dr. Priyanka Khandelwal, Department of Pediatrics, AIIMS Delhi.
     E-mail: drpriyanka8588@gmail.com
   - Dr. Aliza Mittal, Department of Pediatrics, AIIMS Jodhpur, Rajasthan.
     E-mail: alizamittal@gmail.com
   - Dr. Sriram Krishnamurthy, Department of Pediatrics, JIPMER, Puducherry.
     E-mail: drsriramk@yahoo.com
   - Dr. Kirti Sen, Department of Pediatrics, Chacha Nehru Bal Chikitsalaya, Delhi.
     E-mail: kirtisen@gmail.com