**ORIGINAL RESEARCH PAPER**

**SMITH LEMLI OPITZ SYNDROME TYPE 2 ASSOCIATED WITH SEVERE LARYNGOMALACIA, TRACHEOESOPHAGEAL FISTULA AND PYLORIC STENOSIS: A CASE REPORT.**

**Abstract**
Smith Lemli Opitz syndrome is a common inborn error affecting cholesterol synthesis. Association with tracheoesophageal fistula, laryngomalacia and pyloric stenosis together has not been found in literature. It is a treatable genetic defect; a brief introduction on management has been described in this report.

**References.**

**Key Words:** Smith Lemli Opitz syndrome, Tracheo-esophageal Fistula, Pyloric stenosis.

**Abbreviations**
- TEF: Tracheo-esophageal Fistula
- DHC: Dehydrocholesterol

**Introduction**
Smith Lemli Opitz syndrome was first described by David Smith, Luc Lemli and John Opitz in 1964.1 It is a common autosomal recessive type of genetic defect with incidence of 1/10000 to 1/60000.2 It is an inborn error affecting cholesterol biosynthesis with an inherited mutation on DHCR7 gene located on 11q13 chromosome.3 DHRC (dehydrocholesterol reductase) deficiency impairs cholesterol production resulting in increased 7 DHC (dehydrocholesterol) level and decreased cholesterol synthesis which causes developmental dysmorphology. There is wide clinical variation ranging from mild to severe lethal form of syndrome involving multiple internal organs. Severe form (type 2)4 is a rare manifestation, here we are reporting a case of Smith Lemli Opitz type 2 syndrome with uncommon association of pyloric stenosis and tracheo-esophageal fistula. TEF, Laryngomalacia and Pyloric stenosis together in Smith Lemli Opitz Syndrome is a rare manifestation and is not reported in literature to the best of our knowledge.

**Case**
Our case was a 3 month old female baby, delivered at private hospital, referred to us on day 2 of life in view of persistent respiratory distress and stridor. Parents noted noisy breathing, drooling of milk, cough and vomiting while feeding, symptoms were increasing progressively. At 30 days of life baby developed respiratory distress, lethargy and poor feeding for which he was taken to hospital where CPAP was given though symptoms were not improving and child referred to us. On arrival to us, respiratory distress and inspiratory stridor was present, facial dysmorphology noted (Image 1) as dolicocephalic skull, open anterior fontanels, proptosis, bilateral low set ears, micro and retrognathia, high arched palate and syndactyly of left lower 2 and 3 finger (Image 2). On examining abdomen palpable mass felt in epigastrium. With above phenotypic characteristic (*specific to syndrome) we kept diagnosis of Smith lemli opitz syndrome. Investigations revealed hypcholesterolemia, USG Abdomen revealed pyloric stenosis, chest x ray were showing opacities, flexible bronchoscopy was done revealed severe laryngomalacia, tracheomalacia and esophageal fistula. (Image 3 and 4)

**Discussion**
Cases has been reported due to increased awareness and diagnostic availability. Milder phenotypes are common, however severe type 2 is rare and association with tracheoesophageal fistula has not been described. Diagnosis can be made by typical characteristics (* described in case, screening can be done with serum 7 DHC level which was increased. Increase in 7DHC/cholesterol ratio in amniotic fluid or chorionic villi sample can be used for prenatal diagnosis as early as 11–12 weeks.5 It is a treatable genetic defect, since there is decreased cholesterol synthesis (product) which is required in adrenals for steroids synthesis, supplementation with steroids has been seen with positive outcome. HMG Co A inhibitor has role to stop 7 DHC (precursor) formation6. Dietary cholesterol supplements, fat soluble vitamins supplementation have been seen to have beneficial role7.

**Images**
- Image 1. Patient with dysmorphology
- Image 2. Syndactyly.