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RESEARCH ARTICLE

A RARE CASE OF NOSOCOMIAL INFECTION WITH CORONA VIRUS OC43 IN RHIZOMELIC CHONDRODYSPLASIA PUNCTATA TYPE1.

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Abstract

Rhizomelic chondrodysplasia punctata type 1 is an inherited disease with extremely rare presentation. It affects 1 in 100,000 individuals [2]. Rhizomelic chondrodysplasia punctata is associated with significantly delayed development and severe mental retardation. Most children with this condition do not achieve developmental milestones and has growth retardation. RCDP is an autosomal-recessive disease caused by mutations in the PEX7 gene. In this article I presented a rare case of patient with Rhizomelic chondrodysplasia punctata type 1 admitted with pneumonia then he has been infected by nosocomial infection with corona virus and RSV that necessitate shifting him to PICU. At the end due to the severity of respiratory problems of individuals with Rhizomelic chondrodysplasia it becomes the corona virus more dangerous and aggressive then we need special caution and high rate of care from both therapeutic and preventive standpoints.

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Introduction:-

Rhizomelic chondrodysplasia punctata type 1 is an inherited disease with extremely rare presentation. It affects 1 in 100,000 individuals [2]. RCDP is an autosomal-recessive disease caused by mutations in the PEX7 gene. An individual who inherits one copy of a PEX7 gene mutation is a "carrier" and do not have related health problems. An individual who inherits mutations from each parent, is expected to be affected with RCDP. Thus it shows that RCDP is an inherited disease but results only when the genes are inherited from both the parents.

If mother and father both are carriers, the chances of getting affected for a child is 25% in each pregnancy; therefore, it is of utmost importance that the reproductive partner of a carrier be offered testing. A negative does not eliminate the possibility of inheritance of the gene mutation by the child. Rhizomelic chondrodysplasia punctata is associated with significantly delayed development and severe mental retardation. Most children with this condition do not achieve developmental milestones and has growth retardation. These children may also have seizure episodes. It is generally associated with recurrent respiratory infections and breathing problems which may be life threatening. Because of their severe health problems, most children with this do not survive long. It is rare for affected child to live past age 10. The milder form of this disease is characterized milder degrees of rhizomelia along with growth and mental retardation. Distinctive facial features such as prominent forehead, widely set eyes, sunken appearance of face, small nose. Additionally, almost all affected individuals present with cataract. The cataracts are apparent at birth (congenital) or develop in early infancy like in our patient.

Case Report:-

This is a 10 months old boy, presented to ED due to cough and excessive secretion for 2 days. He is a known case of Rhizomelic type chondrodysplasia punctata type 1, History of Left kidney grade 1 hydronephrosis, Left inguinal hernia, moderate ASD and small PDA, Bilateral congenital cataract status post Bilateral lens aspiration and anterior vitrectomy, Gastroesophageal reflux disease on ranitidine, On Gastrostomy feeding tube since birth. He was in his usual state of health until 2 days before He presented to ED with cough that awakes him from sleeping associated with excessive secretion, Shortness of breath and posttussive vomiting twice a day, He had history of contact with sick person. He had history of previous admission in PICU 3 month ago intubated and connected to mechanical ventilation due to viral bronchiolitis for 1 month.

No history of fever, Upper respiratory tract infection symptoms, cyanosis or apnea, or change bowel habit, No history of abnormal urinary symptom, No history of abnormal movement, No history of skin rash.

Past History:

Antenatal and Birth history: Uneventful pregnancy, product of NSVD, admission to ICU due to chondrodysplasia for investigation put on nasal cannula, stay for 2 month +12 day

Immunization: received only birth vaccine

Developmental History: developmental milestones of 1 month old, gross motor: turns head side to side. Speech: cries, social: calms when comfortable.

Family History: + consanguinity, father he is soldier, healthy, mother she is house wife, healthy, 3 sibling 1st one girl, 5 years old healthy has rhizomelic type chondrodysplasia punctata type 1, 2nd one girl 3 years old healthy, The patient is the 3rd one, No hx of chronic disease, hereditary or genetic run in family.

Allergies: No history of allergies

General examination of the patient:

Patient looks well, in moderate Respiratory distress, sub and inter costal retraction with large amount of loose secretions, well perfusion and hydration.

V/S: SBP(mmHg) 101-98

DBP(mmHg) 67-69

HR(Freq./min) 143-143

Temp 37.4-37.3

RR(Freq./min) 38-35

SPO2(%) 82-89-98 on 2L O2 NC

Chest: equal bilateral air entry, bilateral transmitted sound and prolongation of the expiratory phase and wheezes.

CVS: S1 + S2 +0, warm extremities. CRT < 2 sec.

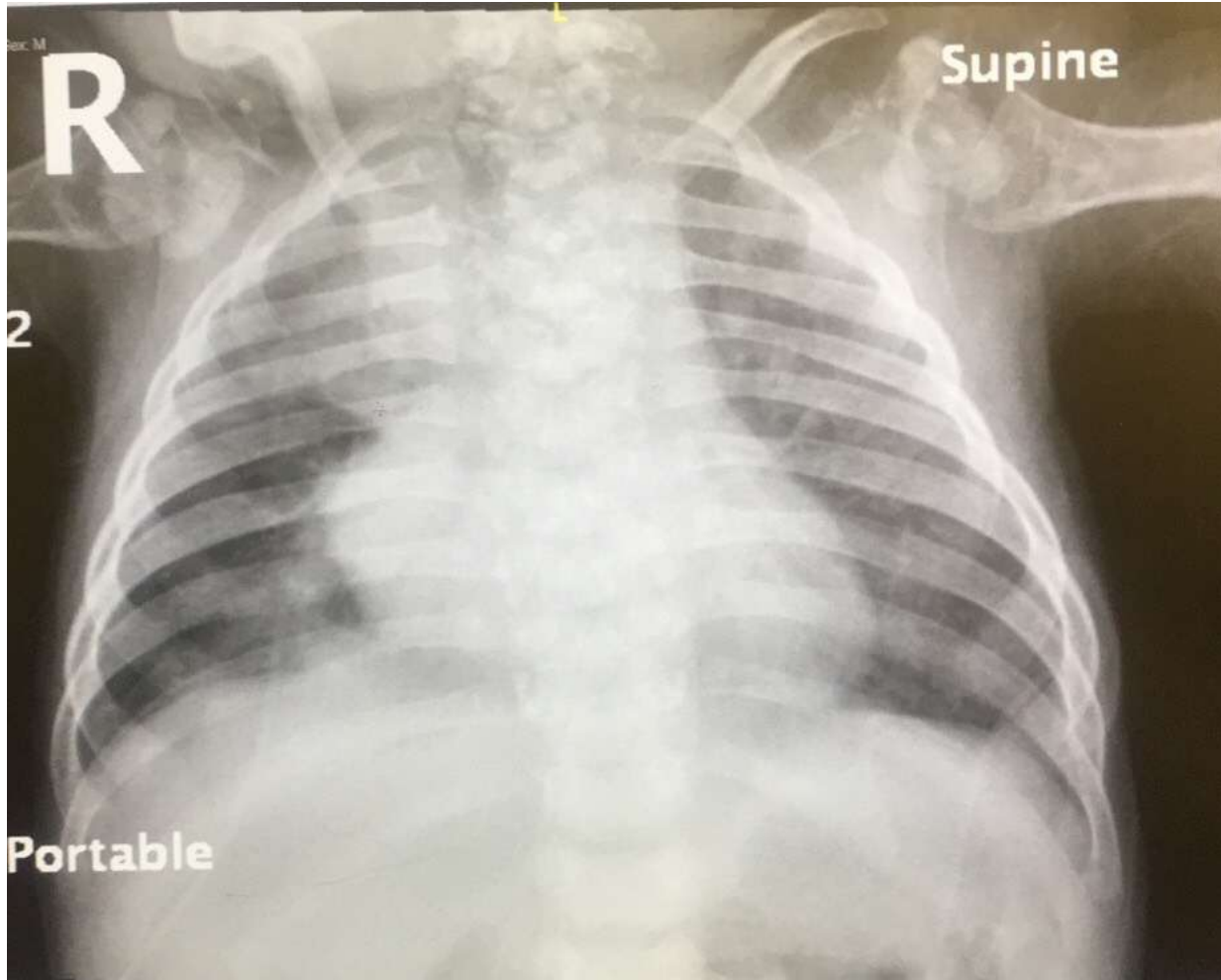
Abdomen: soft and lax, reducible left inguinal hernia, no signs of strangulation.

Musculoskeletal: short arms and legs. contraction of elbow and knee joints, no skin rash.

ENT: congested throat no exudates.

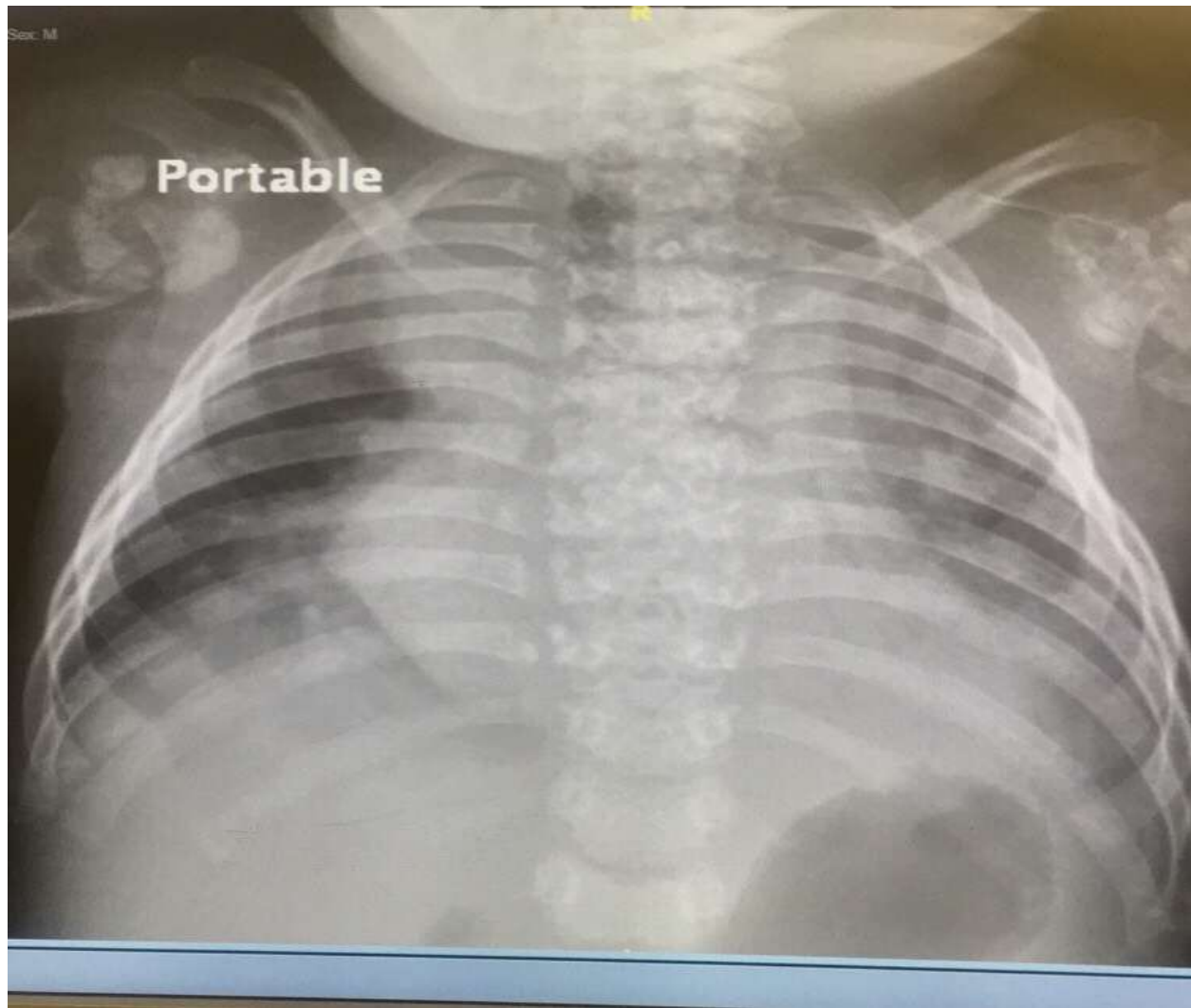
ER COURSE:

He was in moderate Respiratory distress (sub and inter costal retraction), desaturating and fair air entry bilateral with transmitted sound and crepitation. Given salbutamol 3 back to back with minimal improvement and was started on cefuroxime and oxygen 2 L/MIN weaned to 1 L/MIN then 0.5L/MIN.



Chest x-ray showed : Bilateral peribronchial wall thickening related to small airway disease , associated with irregular opacity seen at the right lung lower zone, possibly due to early pneumonia. The cardiomediastinal silhouette is unremarkable . No pleural effusion or pneumithorax ,No interval change.

Then he had been shifted to the ward as case of pneumonia, He was on IV fluid maintenance ,hypertonic solution q6hr , cefuroxime , acetaminophen drops PRN , salbutamol q6hr PRN. After that he had been investigated by nasopharyngeal aspirate specimen and Chest x-ray. coronavirus OC43 and RSV was detected on nasopharyngeal aspirate specimen .



Chest x-ray showed: interval worsening of lung base aeration, more on the left side . the rest of the finding appear unchanged.

Patient showed signs of Respiratory distress with multiple desating down to 40s. He had one episode of apnea observed at night, relived with stimulation, face mask and in some occasion with suctioning. That necessitate shifting him to PICU .He required 2L OXYGEN through Nasal cannula and salbutamol. patient was on clindamycin and ceftriaxone.

Laboratory investigations:

Complete blood count: WBC 17.73

RBC 4.25

Hb 114

HCT 0.360

MCH 26.8

MCHC 319

MCV 83.9

RDW 17.7

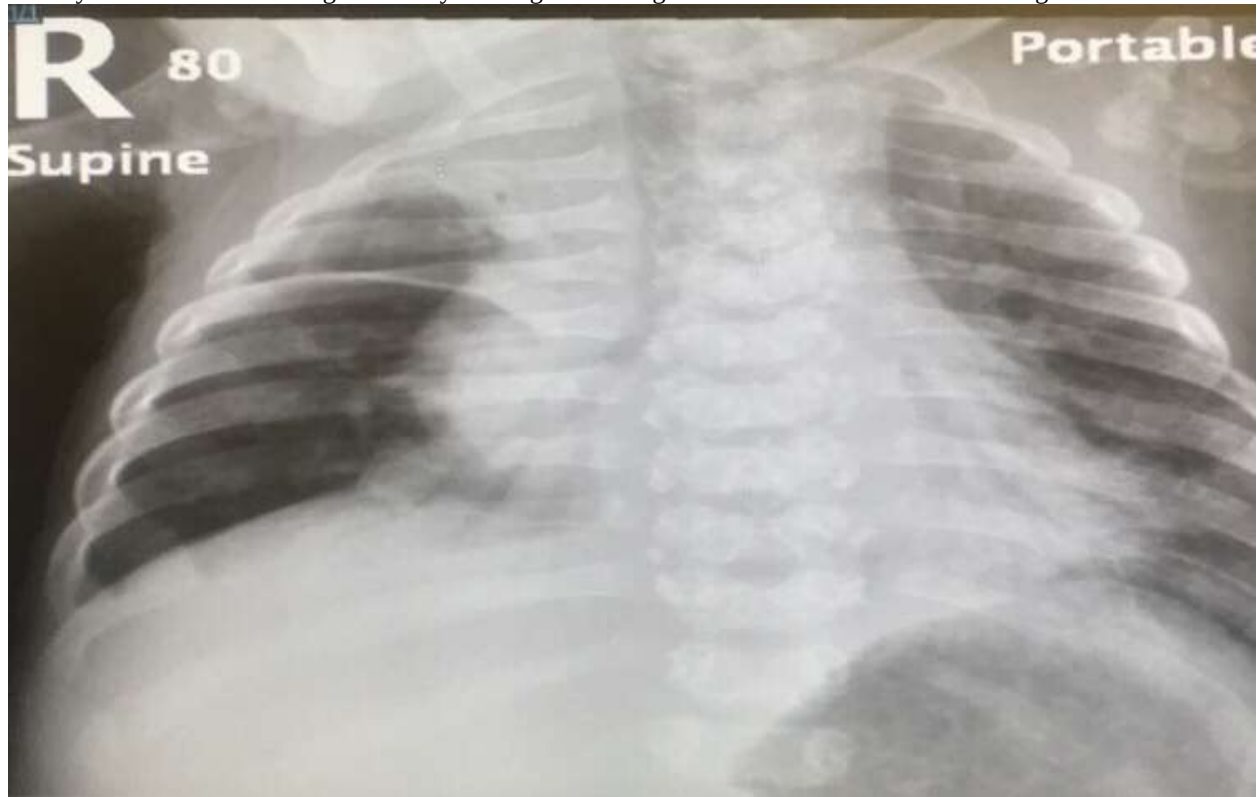
PLATELETS 211

Electrolytes , UR, CR: Sodium 140
Potassium 5.8

Chloride 106
Urea 2.6
Creatinine 33

The last CXR showed:

- mildly better aerated left lung and newly seen right mid lung atelectasis . no other interval change



Discussion:-

Human coronaviruses are known causes of the common cold. Subtype OC43 (HCoV-OC43) is the more prevalent human coronavirus in several parts of the world. Recent studies have suggested these viruses can cause severe lower respiratory tract illnesses in children , but Due to the severity of respiratory problems of individuals with Rhizomelic chondrodysplasia punctata might indicate the bad prognosis and a major cause of morbidity and mortality when he infected with this virus. Because of that they are needs to special caution and high rate of care from both therapeutic and preventive standpoints. And when worsening the patent condition like in our patient we shifted him to PICU to received a proper care.

Conclusion:-

Due to the severity of respiratory problems individuals with RCDP require the influenza vaccine and RSV monoclonal antibody. There has been no systematic study for oral plasmalogen supplementation to determine clinical benefits.

RCDP is a rare presentation in paediatric age group. There is no cure for RCDP. Unfortunately, most children do not survive after age of 10. Diagnosis was done mainly on the basis of clinical examination and investigations such as X-rays. We concluded that, Management of RCDP is symptomatic and supportive and may include physiotherapy and orthopedic procedures (in later stages) to improve function. The child may also undergo cataract surgery to improve vision.

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