inheritance. CGD is characterized by neutrophils and monocytes capable of normal chemotaxis, ingestion and degranulation, but unable to kill catalase-positive microorganisms due to defects in one of the 5 major subunits of NADPH oxidase.

Method and material: The medical records of 38 patients diagnosed with CGD were reviewed and analysed with respect to demographic data, age at presentation and diagnosis, clinical features, laboratory investigations, organisms isolated, treatment & prophylaxis given and clinical course.

Results: Our study had 28 males and 10 females with 13 having history of consanguinity. Their mean age at presentation and diagnosis was 1.32yr and 2.5yr respectively. The most common manifestations at presentation were failure to thrive (100%) and lymphadenopathy (100%) followed by hepatomegaly (72%) and splenomegaly (48%). The commonest infection was pneumonia (84%) followed by abscesses (55%) involving lungs, liver, subcutaneous tissue; osteomyelitis (15%); urinary tract infections; otitis media and CNS infections. Organisms isolated from blood, stool and pus of infected lesions included bacteria- Mycobacterium tuberculae (50%), Staphylococci (24%), Klebsiella (16%) and fungi- Aspergillus (13%), Candida (26%). Biopsies done in 36% patients from lymph node, skin, lung and liver showed non caseating granulomatous inflammation. Diagnosis was based on reduced nitroblue tetrazolium test (NBT) between 0-5% in all patients and confirmed by dihydrorhodamine (DHR) assay in 84% patients. 18 families were screened. All patients received antibiotics, 80% received AKT, 76% received Antifungals and all received antifungal and antibacterial prophylaxis. 4 patients have succumbed to the illness and 13 patients are lost to follow-up. 7 patients inherited CGD in an X-linked recessive fashion. Genetic mutation analysis has been done in 22 patients.

Conclusion: CGD is a not uncommon in India. The commonest mode of presentation was Pneumonia, skin and subcutaneous abscesses, lymphadenitis and osteomyelitis. Mycobacteria, Staphylococcus Aureus, Klebsiella, Aspergillus, Candida, and Gram negative bacilli were the commonest organism isolated in our series. Infection with Aspergillus, Burkholderia Cepacia, Serratia Marcescens, Nocardia should prompt work up for CGD. All children with CGD should be given routine chemoprophylaxis with Septran and Itraconazole. Families should be screened and counselled during future conceptions.

PID_ALP_ID-1_V1.4
AN INTERESTING CASE OF FEVER-NEONATAL ONSET MULTI SYSTEMIC INFLAMMATORY DISORDER

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Background: Neonatal onset multisystem inflammatory disease (NOMID) is the most severe phenotype in spectrum of Cryopyrin (NLRP3/NALP3) associated periodic syndromes (CAPS) associated with chronic IL-1B over production. Mutated NLRP3 causes constitutive activation of NLRP3 Inflammasome which over produces IL-1B. The role of IL1 B in NOMID has been demonstrated in clinical trials using IL1 blocking agents that cause rapid resolution of disease manifestations.[1]

Case Summary: A 16 months old male child presented with complaints of fever and rash since day 1 of life and delayed development. Fever is almost always associated with rash. Rash is urticarial, truncal with itching. At 7 months child was noticed to have anemia with hepatosplenomegaly and neuroimaging was s/o mild cerebral atrophy with communicating hydrocephalus. Patient had h/o 3-4 episodes of pneumonia too. Examination revealed overhanging forehead, hypertelorism, depressed nasal bridge. Child had pallor, rash, hepatosplenomegaly with generalized hypotonia. A differential of primary immune deficiency disease v/s inflammatory disorder v/s mastocytosis v/s storage disorder was evaluated. His baseline investigations were within normal range. MRI was s/o mild generalized atrophy with dilated lateral ventricles. Bone marrow aspiration was done to r/o mastocytosis. Liver biopsy had no evidence of storage disease. Lymphocyte subset and DNT cell analyses were within normal range. After ruling out the above differentials a periodic inflammatory syndrome was suspected. Cryopyrin mutation analysis at NIH suggested mutation in CIAS1 G755R region. This mutation is interesting as it is exon 4, which is not a common mutation.

Discussion: A basic innate response to any pathogen is Inflammasome formation.[2] In CAPS, mutated NALP3 is constitutively activated resulting in increased assembly of NALP3 infalmmasome and active caspase 1. Active caspase 1 cleaves inactive pro IL 1 B to its active form. NOMID has onset in neonatal or early infancy. It has continuous flares with involvement of skin (rash), eye (conjunctivitis), joints, ears, and meninges.[1] Distinguishing features being chronic meningitis, SNHL and a characteristic facies. IL 1 receptor antibodies (Anakinra) are used in the treatment. With treatment, systemic inflammation, conjunctivitis, arthritis and cochlietis can be prevented. Leptomeningitis can be fully reversible.

Conclusion: NOMID is a CAPS presenting with fever since early life. The cardinal features are fever with rash since day 1 of life with continuous flares. The present case of NOMID had an exon 4, rather than exon 3 where most of the identified mutations are seen. After differentiating it from immunodeficiency disorder and since the pathophysiology of NOMID involves IL-1 B over production, IL1 antibodies are used in treatment, which cause prevention of end organ damage.

PID_ALP_ID-1_V1.6
TOPIC: LEUKOCYTE ADHESION DEFICIENCY TYPE 1 – A TERTIARY CARE CENTRE EXPERIENCE

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Introduction: Leukocyte adhesion defect (LAD) is a rare, autosomal recessive primary immunodeficiency disorder of phagocytes with defective aggregation at the site of infection due to the absence of surface integrins. Diagnosis is based primarily on flow cytometric analysis of neutrophils for the surface expression of CD11, CD18 and CD15s. More than 300 cases have been described for LAD 1 worldwide, while for LAD 2 and LAD 3; there are less than 10 cases each.

Materials and methods: Analysis of clinical and laboratory profile of 6 cases of LAD type 1 seen at our institute over a period of 5 years (2011-2016) Demographic data was collected with respect age, consanguinity, community and residence. Taking an average of WBC count on presentation, mean WBC count was calculated. Microorganisms identified from respective in situ sites were documented. The CD 11, CD18 and CD15s expression identified with flow cytometry analysis. The data obtained being tabulated and expressed in graphical form.

Results: The age of presentation was from 1 to 6 months. Five were born of consanguineous marriage. 4 patients were from Maharashtra and 2 from Gujarat. 3 patients belonged to Hindu Maratha, 2 to Muslim Sunni and one to Hindu Kathiawadii community. All had delayed separation of umbilical cord on questioning. Average neutrophil count at presentation was 67,788/mm^3. Of the 6 cases, 4 had gram negative septicemia. Pseudomonas was isolated from blood culture in two cases and ET culture in one. In one patient, Klebsiella was isolated in urine and enterococci in blood. Axillary and inguinal ulcers were seen in one and necrotizing otitis externa in the another patient. All cases were severe LAD type 1 with CD 18 and CD11 a, b and c ranging from 0% in 4 and 1.6% in one. 3 patients expired from septicemia and one was lost to follow up. One case is hospitalized with necrotizing otitis externa with LMN facial palsy. One case is posted for transplantation at AIIMS, New Delhi.

Conclusion: LAD is a rare disease characterized by lack of cell surface expression of integrins, which are essential for adhesion of leukocytes to endothelial cells and chemotaxis. LAD-1 caused by lack of CD-18 integrins on the neutrophil surface has the worst prognosis and most patients die within 1st year due to severe infections;< 1% expression are clinically severe, whereas those with 2.5-10% expression are moderate to mild. In LAD-II, the neutrophils lack the membrane carbohydrate Sialyl-Lewis X required for adherence to activated endothelial cells. LAD-III is associated with defect in activation of Rap 1 protein.

LAD-I is characterized by delayed shedding of umbilical stump, recurrent bacterial infections of the skin, leucocytosis, absence of pus and poor wound healing. All patients in present study had delayed separation > 10 days (Mean 5.8-10.9). Commonest organisms isolated are Staphylococcus aureus and enteric gram negative bacilli and in our study pseudomonas was in majority.