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Date: 29/09/2026 01:29 PM
Subject: CPC Clinical handout. 30.09.2026

Dear All,

The next Wednesday CPC of the academic session will be held tomorrow, **September 30, 2026** at **08.00 hours** (IST) in **Lecture Theatre 1**, Nehru Hospital, PGIMER, Chandigarh under the Chairmanship of **Prof. Sanjay Jain**.

The session will also be available on the Webex platform. Kindly follow the link below to join.

<https://pgitelemed.webex.com/pgitelemed/j.php?MTID=md00288711fdbf94321a0819943e426a2>

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The Clinical handout of the case to be discussed is attached herewith.

The clinical aspect will be discussed by **Dr. Vignesh P, Department of Pediatrics**. Radiology will be discussed by **Dr. Anmol Bhatia** and the autopsy pathology will be presented by **Dr. Hemlata**.

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Regional Resource Centre, North
Department of Telemedicine
PGIMER, Chandigarh

Staff CPC (30/09/2026)- Clinical Protocol

AG, 4.5 years; Male; R/O Burdwan, West Bengal
CR No. 20**-02**-**15
DOA: 26.07.2026 15:14 HRS
DOD: 26.07.2026 19:50 HRS

Unit In-charge: Allergy Immunology/ Emergency
Clinical discussant : Dr. Vignesh P
Radiology discussant : Dr. Anmol Bhatia
Pathology discussant : Dr. Hemlata

Second-born child to a third-degree consanguineously married couple was symptomatic from the age of 2.5 months in form of multiple joint swelling. Illness started with swelling in right little toe, followed by involvement of right knee, both wrists, left knee, and bilateral ankles. He had fever with maculopapular rash at age of 4 months. He also developed localized BCGitis at 2.5 months of age that persisted until 6 months of age.

MRI left knee (at Kolkata) revealed effusion and synovial thickening. He was initially managed as a case of juvenile idiopathic arthritis and received intra-articular steroid injections to left knee. At 6 months of age, he developed loose stools and was evaluated at a tertiary care centre at Bengaluru, found to have low IgG levels (145 g/dL), and was commenced on replacement intravenous immunoglobulin therapy.

History of one male sibling loss at the age of 7 months who had history of recurrent pneumonia, dactylitis and oral thrush. Received all vaccines up to 14 weeks of age including OPV and rotavirus vaccine.

He presented at the age of 1.5 years with persistent diarrhea and failure to thrive.

1st admission in PGI (06th to 13th May 2023): Child was noted to have severe malnutrition, pallor, abnormal looking BCG scar, hepatosplenomegaly, dactylitis, and deforming polyarthritis. Basic investigations revealed anemia, transaminitis, normal ESR and CRP. CMV work-up was negative. Immunological and molecular investigations confirmed MHC class 2 defect with pathogenic homozygous missense variant in *RFXANK*. Started on nutritional supplements, anti-microbial prophylaxis (cotrimoxazole and itraconazole) and modified anti-tubercular therapy (ethambutol, levofloxacin, and azithromycin) for BCG disease. High-resolution HLA typing was sent for the child and parents.

3 OPD visits (Aug 2023 to Sep 2024): A short course of oral prednisolone was tried for polyarthritis with minimal response; subsequently had oral thrush requiring oral fluconazole and an episode of otitis media. Intermittent episodes of loose stools re-appeared. Stool parasitic work-up was non-contributory. Received empirical course of nitazoxanide. Received 3 PRBC transfusions for anemia between the duration January and April 2025.

2nd admission in PGI (13th Apr to 07th May 2025): Child was admitted for chronic diarrhea, oral thrush, and weight loss of 3 kg. Stool PCR showed positivity for Norovirus G2. Electrolyte disturbances – hypophosphatemia and hypokalemia were managed, and nutritional rehabilitation measures were initiated. IV albumin 1 g/kg was given in view of severe hypoalbuminemia and pedal edema. High dose IVIg and oral ribavirin were tried for norovirus; however, ribavirin later withheld due to worsening transaminitis. Oral administration of gamma globulin 2.5 g/day was given for 6 days that resulted in improvement of symptoms.

Multiple OPD visits (May to Nov 2025): One episode of anemia requiring PRBC transfusion; flare of arthritis requiring oral prednisolone; intermittent episodes of loose stools persisted. Developed extensive dental caries and re-occurrence of bilateral ear discharge was also noted. ENT evaluation revealed bilateral tympanic membrane perforation. Oral immunoglobulin therapy was tried intermittently for norovirus diarrhea with moderate benefit. Oral sirolimus was also added.

3rd admission in PGI (26th Nov to 05th Dec 2025): Admitted for loose stools, oral thrush, and weight loss. Weight: 8.7 kg (-5.19 Z), Height: 82 cm (-5.12 Z), W/H: -2.75 Z. Stool PCR revealed Norovirus G2. One-unit PRBC transfused for anemia. Purge rate improved with oral immunoglobulin therapy. Oral nitazoxanide was also added.

3 OPD visits (July 2026): Intermittent loose stools and gaseous abdominal distension; transaminitis resurfaced. Oral gamma-globulin therapy tried for persistent loose stools. A PRBC transfusion given for anemia.

Both parents developed viral conjunctivitis and 5 days later, child developed 2 episodes of vomiting followed by respiratory difficulty and was admitted in emergency the next day.

Weight: 8 kg (-5.4 Z); **HR:** 124/min; **RR:** 60/min; **BP:** 96/66 mm Hg; feeble peripheral pulses; **SpO2:** 100% RA

Examination:

Cardiovascular: Unremarkable

Respiratory system: No added sounds

Abdomen: Distended, liver 3 cm below RCM. Spleen 2 cm below LCM.

CNS: E4V3M6

Investigations:

Blood counts	06/05/23	14/04/25	10/05/25	22/11/25	07/07/26	17/07/26	27/07/26
TLC (per mm ³)	18,800	5,600	9,090	5,900	6,300	4,500	10,000
Differential counts	P ₂₇ L ₅₂ M ₁₁ E ₀₉	P ₃₂ L ₅₀ M ₁₅ E ₀₁	P ₄₁ L ₃₆ M ₁₅ E ₀₆	P ₅₀ L ₄₁ M ₀₇ E ₀₁	P ₄₄ L ₃₅ M ₁₆ E ₅	P ₄₈ L ₂₆ M ₂₃ E ₀₂ B ₀₁	P ₈₅ L ₀₆ M ₀₈ E ₀₁
Hb (g/dL)	9	8.2	6.4	7.4	6.4	10.8	8.7
MCV (fL)		83.5	-	59	61	67.8	73.2
Platelets	5.45 L	2.58 L	1.73 L	2.43 L	2.55 L	2.73 L	3.29 L

Biochemistry	06/05/23	14/04/25	10/05/25	22/11/25	17/07/26	26/07/26	26/07/26
Na ⁺ / K ⁺	137/ 5.0	135/ 3.65	-	131/ 3.2	132/ 3.9	129/ 4.5	127/ 6.15
Urea / Cr	32/ 0.22	11/ 0.27	29/ 0.3	5/ 0.22	08/ 0.27	08/ 0.41	30/ 0.89
T proteins/ Alb	5.9/ 3.9	6.1/ 3.2	-	5.9/ 3.6	7.0/ 4.0	5.1/ 3.0	5.7/ 2.8
TB/ CB	0.9/ 0.34	0.59/ 0.15	1.36/ 0.44	0.7/ 0.1	0.46/ 0.1	0.53/ 0.12	1.43/ 0.72
AST	611	55	452	71	212	202	345
ALT	374	37	307	46	130	97	95
ALP	204	84	266	91	120	99	153
Cal/ Phos	9.4/ 5.5	8.4/ 3.3		7.7/ 1.8	8.6/ 2.1	7.0/ 5.1	5.7/ 2.8
CRP	0.23	0.15	1.08	13.8	6.15	1.77	4.19

Microbiology	Date	Result
Blood Culture (BACTEC)	26/07/2026	<i>Acinetobacter baumannii</i> : IS to Minocycline, R – others <i>Enterococcus faecium</i> : S to Teicoplanin, gentamycin, vancomycin Minocycline
Fungal Culture	26/07/2026	No growth of pathogenic fungus
HCV IgM serology	20/07/2026	Non-Reactive
HBsAg	20/07/2026	Non-Reactive

VBG	26/07/26
pH	7.18
pCO ₂	16.8
SO ₂	85.5%
cHCO ₃	6
cLac	11.5

Hb electrophoresis (2022):
HbF-8.5%; HbA-86.9%;
HbA2-5.1%

Stool multiplex virus PCR (16/04/24, 05/04/25, 15/11/25): Positive for Norovirus G2, negative for norovirus G1, rotavirus, adenovirus, astrovirus, and sapovirus

Stool multiplex virus PCR (July 2026): Negative for all above viruses

Blood CMV viral load (15/05/23): negative; HIV serology: Negative

Date	22/07/22
IgG	1.45 (4.10-10.81) g/L
IgA	0.26 (0.13-0.82) g/L
IgM	0.35 (0.48-2.28) g/L

Lymphocyte subsets (May'23)	CD3- 45.8% (39-73); CD19- 12.97% (6-23); CD56- 43.16% (3-22)
T lymphocyte subsets (May'23)	CD4+T- 47.49% (32-51); CD8+T- 26.3% (14-30); Normal CD4:CD8 ratio; decreased proportion of naïve T cells
T lymphocyte proliferation (May'23)	Normal proliferation in response to PHA; slightly reduced with CD3/CD28
HLA-DR in B lymphocytes (May'23)	0% in the patient compared to 13.4% in healthy control

Invitae PID gene panel: Homozygous variant in *RFXANK* c.380C>G (p.Pro127Arg)

Sanger Sequencing: Parents are heterozygous carriers for the same variant

Course and management: Child was admitted in emergency with grunting and shock. He was started on IV meropenem and oseltamivir. IV dextrose bolus given in view of hypoglycemia (RBS- 65). He received normal saline bolus of 10ml/kg for shock followed by maintenance fluids. He developed cardiac arrest and succumbed to his illness.

Unit's final diagnosis: MHC class 2 defect (RFXANK-related)/ Beta-thalassemia trait with chronic norovirus diarrhea and malnutrition, multi-system viral illness, bacterial sepsis, and shock.