with Mycobacterium Avium Complex), DCML (dendritic cell monocyte and lymphoid deficiency) and Emberger syndrome (sensorineural deafness, congenital lymphedema and viral warts). This is a sporadic entity with autosomal dominant inheritance. Germline *GATA 2* mutations are the most common defect predisposing to pediatric myelodysplastic syndrome with a high prevalence of monosomy 7 thus mandating its evaluation in every case of monosomy 7 associated MDS [3]. Allogeneic hematopoietic stem cell transplantation is the only curative option for both immunodeficiency as well as MDS/AML [4-6].

Evaluating the cause of repeated infections without dismissing them as related to malignancy/chemotherapy has helped us in reaching a diagnosis. Identification of such genetic predisposition not only helps us manage our patients better, but also has implications for donor search for bone marrow transplantation and genetic counseling for the family.

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Acquired Recto-Vaginal Fistula as a Presenting Feature in an Infant with Severe Combined Immunodeficiency

Acquired recto-vaginal fistula in infancy is a rare entity. We report a child with Severe Combined Immunodeficiency (SCID) who presented to us with an acquired recto-vaginal fistula.

A 5½-month-old girl, firstborn of a non-consanguineous marriage presented with constipation for one month and passing stools per vaginum for three days. She was born at full term at our centre, with a birthweight of 3100 grams, to HIV negative parents. Complete physical examination, including the perineum was noted to be normal at birth. She had been hospitalised for culture-negative sepsis without meningitis at day 21 of life and uncomplicated dengue fever at three months of age. She was on exclusive breastfeeding and had received BCG, OPV, DPT, and Hepatitis B vaccines with no reactions. There was no family history of immunodeficiency disorders or unexplained infant deaths. There was no history of trauma, surgery or suspicion of abuse. Her weight was 6.4 kg (0 z score), length was 59 cm (0 to -2 z score). She had a typical BCG scar. Examination of the perineum revealed a fistulous opening next to the vaginal orifice at the nine o'clock position, suggestive of a recto-vaginal fistula and there was no obvious evidence of local infection. Systemic examination was otherwise unremarkable.

Hemogram showed a normal absolute lymphocyte count (ALC) of 7385 cells/cu.mm. She underwent a diversion colostomy, and per-operatively was found to have a fistulous tract extending from the vagina to the rectum by gentle probing. There was no gross evidence of

inflammation or infection. On post-surgery day 3, she developed a new-onset fever along with multiple nodular lesions all over the body and received intravenous piperacillin-tazobactam and vancomycin for presumed bacterial sepsis. On day 10, she developed a burst abdomen and underwent a surgical closure. She was suspected to have an underlying predisposing condition like primary immunodeficiency or inflammatory bowel disease. Biopsy of the skin nodule showed panniculitis suggestive of erythema nodosum. Xpert MTB/RIF assay of the tissue was positive for *Mycobacterium tuberculosis* (MTB) complex with no rifampicin resistance. HIV ELISA and NBT (Nitro blue tetrazolium) test were negative. Flow cytometry was suggestive of T-B+NK- type of SCID [$CD4^+$ T cell count - 43 cells × 10^9/l (9700 - 2200 cells × 10^9/l), $CD19^+$ B cell count- 2062 cells × 10^9/l (390-1400 cells × 10^9/l) and $CD3^-/CD16^+$ $CD56^+$ NK cell count-41 cells × 10^9/l (130-720 cells × 10^9/l)]. Her serum immunoglobulin levels were deficient [IgG -2348 (4669-10673) µmol/l, IgM-77 (41-237) µmol/l, IgA-176 (438-2500) µmol/l, IgE- 10.9 (0.96-842)µg/l]. Whole exome sequencing revealed a homozygous *JAK 3 kinase* mutation. Ophthalmology examination and echocardiography were normal.

She developed severe progressive pneumonia requiring ventilator support and succumbed despite treatment with anti-tuberculous drugs, antifungals, antibiotics, and co-trimoxazole for a presumed *Pneumocystis* pneumonia. Bacterial, fungal, and mycobacterial cultures of the biopsy tissue, quantitative CMV DNA PCR in blood and *Pneumocystis jirovecii* staining of the endotracheal aspirate were negative. Parents were counselled to undergo genetic screening and prenatal testing for subsequent pregnancies.

Recto-vaginal fistula, a rare type of anorectal malformation with abnormal epithelial lined connections between the rectum and vagina, has an incidence of less than 1% in children [1]. They can be congenital or acquired. Acquired causes include trauma or infections in the recto-vaginal septum such as anorectal abscesses and Bartholin gland infections, tuberculosis and lymphogranuloma venereum [2]. Rarer causes include malignancies, inflammatory bowel disease, post-radiation therapy, and operative traumas. Acquired recto-vaginal fistulas have been reported to occur in infants with HIV from Africa, representing an early manifestation of the disease [3-5]. The postulated pathogenesis is probably, secondary to a low grade and indolent perianal infection [3]. In the previous case series in HIV-positive girls, the fistulae were small and extending anteriorly from the rectum to distal vagina [3]. Most of these children had evidence of tuberculosis or lymphocytic interstitial pneumonia in the lung [3]. However, rectal fistulae in boys with HIV have only been reported rarely [6]. Surgical closure is usually unsuccessful because of poor wound healing [4,6].

SCID is a medical emergency as hematopoietic stem cell transplantation can prevent death in children before infections occur. Unusual presentations like acquired recto-vaginal fistula should be kept in mind to expedite diagnosis and management.

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