**Table 1.**
Canadian Inborn Errors of Immunity National Registry Pilot Data.

| Characteristics | Pediatric Cohort* | Adult Cohort** |
|---|---|---|
| Recruited participants | 20 | 42 |
| **Sex** | n (%) | n (%) |
| Male | 12 (60) | 10 (24) |
| Female | 8 (40) | 32 (76) |
| | Median (range) | Median |
| Age at the time of symptom onset (years) | 1 (0.08–4) | n/a |
| Age at the time of diagnosis (years) | 5 (1–15) | n/a |
| | Mean (SD) | Median (range) |
| Age at last visit (years) | 11 (5.1) | 63 (18–77) |
| **Diagnosis (IUIS classification)** | n (%) | n (%) |
| ***1. Predominantly antibody deficiency*** | ***15 (75)*** | ***30 (71)*** |
| CVID with no gene defect specified | 10 (50) | 17 (40) |
| Selective antibody deficiency (IgA/IgM deficiency) | 1 (5) | 9 (21) |
| X-linked agammaglobulinemia with an associated BTK mutation | 2 (10) | n/a |
| Hypogammaglobulinemia with absent B cells, no genetic evaluation | n/a | 3 (7) |
| TACI deficiency | 2 (10) | n/a |
| APDS | n/a | (2) |
| ***2. Immunodeficiencies affecting cellular and humoral immunity*** | ***n/a*** | ***2 (5)*** |
| Low CD4 count | n/a | 2 (5) |
| ***3. CID with associated syndromic features*** | ***1 (5)*** | ***4 (9.5)*** |
| SPNS2 deficiency | 1 (5) | n/a |
| Hyper-IgE syndrome | n/a | 4 (9.5) |
| ***4. Auto-inflammatory disorders*** | ***1 (5)*** | ***n/a*** |
| ADA2 deficiency | 1 (5) | n/a |
| ***5. Diseases of immune dysregulation*** | ***3 (15)*** | ***n/a*** |
| XMEN | 2 (10) | n/a |
| LRBA deficiency | 1 (5) | n/a |
| ***6. Phenocopies of PID*** | ***n/a*** | ***2 (5)*** |
| Good's syndrome | n/a | 2. (5) |
| ***7. Secondary immune deficiencies*** | ***n/a*** | ***3 (9.5)*** |
| **Immunoglobulin replacement therapy** | n (%) | n (%) |
| IVIG | 9 (45) | 7 (17) |
| SCIG | 11 (55) | 21 (50) |
| **Infections in the last 12 months** | n (%) | n (%) |
| Respiratory tract infections (sinusitis, pneumonia) | 11 (55) | 26 (62) |
| Gastrointestinal infections | 2 (10) | 19 (45) |
| Ear infections | 9 (45) | 5 (12) |
| Skin infections | n/a | 3 (7) |
| Abscess | n/a | 5 (12) |

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ADA2 deficiency- adenosine deaminase 2 deficiency, APDS-activated p110δ syndrome, CID -combined immune deficiency, CVID -common variable immune deficiency, IVIG – intravenous immunoglobulin, LRBA deficiency - lipopolysaccharide-responsive and beige-like anchor protein deficiency, PID -primary immune deficiency, SCIG – subcutaneous immunoglobulin, SPNS2 deficiency- Spinster homolog 2 deficiency, TACI deficiency - hypogammaglobulinemia due to TNFRSF13B mutation, XMEN-X-linked immunodeficiency and magnesium defect, EBV and neoplasia.

**Conclusion:** The information collected in the CIEINR will be essential to analyze unique features of IEI in Canada and will provide a strategic method to optimize patient care and improve access to novel therapies and clinical trials. Registry data will provide a tool to assess barriers to medical care, particularly for those in rural/remote regions, and will be vital to support advocacy for resource allocation.

**Keywords:** Registry, Inborn Errors of Immunity, Canada

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**(4)**

## A novel BNLK mutation presenting with hepatopathy and rickets

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BLNK represents a central linker protein that bridges the B cell receptor–associated kinases with a multitude of signaling pathways and may regulate the biologic outcomes of B cell function and development. Blnk deficiency is a type of autosomal recessive immune disorder that involves the absence of B cells, agammaglobulinemia, and recurrent infections. Only ten patients with BLNK deficiency have ever been identified.
We present a 29-year-old Turkish female with BLNK deficiency with a novel homozygous CGA > TGA codon123, exon6) mutation in the BLNK gene. She developed severe liver failure and rickets at the age of 12. Although BLNK mutations are rare causes of agammaglobulinemia, it is important to consider them in patients with B cell deficiency.
She is the first to be described with hepatopathy and bone metabolism disorder in BLNK deficiency in the literature. Hepatopathy and rickets, seen in this case, are clinical conditions that should be considered in BLNK deficiency.

**Keywords:** BLNK deficiency, Inborn errors of Immunity, Liver failure, Rickets, Hepatopathy

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