

# Incidence of Primary Immunodeficiency Disorders at a Tertiary Care Immunology Clinic

Abstract #11895

Benjamin Prince MD<sup>1,2</sup>, Kelsey Lecerf MD<sup>1</sup>, Peter Mustillo MD<sup>1,2</sup>, and Rebecca Scherzer MD<sup>1,2</sup>

<sup>1</sup>Pediatric Residency Program; <sup>2</sup>Division of Allergy and Immunology; Nationwide Children's Hospital, Columbus, OH.

## BACKGROUND

### Prevalence of Primary Immunodeficiency Disorders

- Primary Immunodeficiency Disorders (PIDD) used to be thought of as rare diseases but with increased awareness of these disorders as well as advanced diagnostic techniques, the prevalence of PIDD has been increasing.
- Newer studies have placed the prevalence of PIDD as high as 1:2000 in children and 1:1200 in adults<sup>2,4,5</sup>

### Diagnosis of Primary Immunodeficiency Disorders

- Most primary immunodeficiencies are not detected until later in life<sup>3</sup>
- There is often delay in the diagnosis of PIDD and this delay can lead to serious, life-threatening infections and complications<sup>5,6</sup>
- Primary care providers are often the first to evaluate these patients and most patients are subsequently referred on to specialists for diagnosis.

## OBJECTIVE

- To determine the incidence of primary immunodeficiency disorders from a population of patients referred due to concern for PIDD found in our weekly Immunology clinic.
- The clinic is staffed with 4 faculty members, a Genetic Counselor, and Social Work.

## METHODS

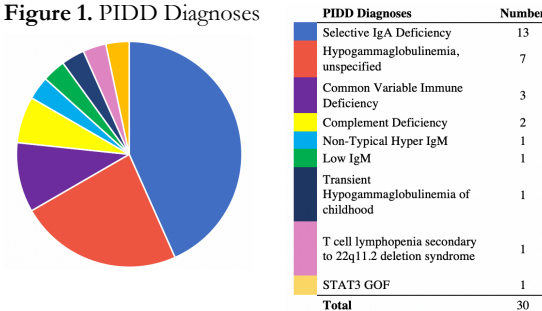
- IRB approval was obtained
- A comprehensive chart review was performed on all patients referred to our Immunology Clinic from January 1<sup>st</sup>, 2018 through December 31<sup>st</sup>, 2018.
- Demographic data collected included the patient's date of birth, age at referral, sex, and ethnicity.
- Additional data collected included referral reason, referring provider, referring provider specialty, length of time between referral and evaluation, subsequent diagnoses, and potential treatment.

## RESULTS

**Table 1.** Demographic Data Of Referred Patients

	Number Referred	Percentage	Number Diagnosed	Percentage
<b>Sex</b>				
Female	84	46	11	37
Male	98	54	19	63
<b>Ethnicity</b>				
White	144	79	22	73
African American	14	8	3	10
Bi-racial	12	7	3	10
African	4	2	1	3
Asian	3	2	0	0
Hispanic	2	1	0	0
Native American	1	<1	0	0
Not Reported	2	1	1	3
<b>Age at Referral</b>				
< 1 year	10	5	0	0
1-3 years	42	23	3	10
4-6 years	32	18	7	23
7-10 years	42	23	7	23
11-17 years	51	28	12	40
18 years and older	5	3	1	3

**Figure 1.** PIDD Diagnoses



**Table 2.** Referrals by Subspecialty Type

Specialist Type	Number of Referrals	Specialist Type	Number of Referrals
Gastroenterology	20	Cardiology	3
Infectious Disease	18	Nephrology	2
Plastic Surgery	16	Allergy and Immunology	1
Pulmonary	16	Neonatology	1
Rheumatology	13	Neurology	1
Endocrinology	9	Neurosurgery	1
ENT	8	Surgery	1
Genetics	8	Developmental and Behavioral Pediatrics	1
Hematology/Oncology	6		

## RESULTS



- Majority were referred by sub-specialists (70%), the most common being Gastroenterologists and Infectious Disease providers.
- The average age at time of referral was 7.6 years.
- Thirty patients (19.3%) were subsequently diagnosed with a PIDD, with selective IgA deficiency being the most common diagnosis (13/30).

## CONCLUSIONS

- Almost 20% of the patients referred to the Immunology clinic were identified as having a PIDD,
- The appropriate diagnosis of PIDD is critical to ensure critical education, patient precautions, and potential treatment
- This relatively high number emphasizes the importance of a timely evaluation and diagnosis, as well as proper education to referring providers about what signs and symptoms to look for when considering primary immunodeficiency disorders.

## REFERENCES

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