Approach To Immunodeficiency

When to suspect an immunodeficiency?

1) Are there are **clinical signs** of immune deficiency?
2) What **arm of the immune system** could be affected?

WARNING SIGNS

SPUR: **s**evere, **p**ersistent, **u**nusual, **r**ecurrent infections +/or autoimmunity

- **> 4 Acute Otitis Media** in 1 year
- **4 twos: > 2** sinus infections; **> 2** pneumonias (CXR proven); **> 2** deep seated infections; **> 2** months on antibiotics
- **4 F's: Furuncle:** recurrent, deep skin/organ abscesses; **Fungus:** persistent thrush after age 1 or recurrent treatment needed; **Failure to thrive,** chronic diarrhea; **Family History:** positive for immune-related issues
- **IV Antibiotics:** required to clear infections

HISTORY

Infection History:

- **Types**: bacterial, viral, fungal
- **Details**: site, onset, frequency, severity
- **Pus production**: yes/no
- **Investigations**: tests done; organism identified
- **Therapy**: tx received & response to tx
- **Wound healing**: normal/delayed
- **Umbilical cord**: delayed separation

Exposures:

- Sick contacts
- Daycare/School
- Travel/Outdoor (Camping/Farming)
- Animals/Pets
- Chemicals/Toxins
- Tobacco, alcohol
- **Vaccinations** (obtain records)

Autoimmune/allergic:

- Cytopenias
- Early onset IBD (<7 yo)
- Lupus
- Joint Pain
- Rashes, eczema
- Asthma

Early severe** presentation, **recalcitrant to Tx

Family History:

- **CONSANGUINITY**
- Immune deficiency
- Early childhood deaths
- Cancer, congenital abnormalities, syndromes
- Autoimmune disease

PHYSICAL EXAM

General:
- **Growth**: normal/delayed
- **Dysmorphic features**: congenital anomalies
- **Hair/nails**: loss of eyebrows, nails

HEENT:
- **Ear issues**: OM, TM scarring, ear anomalies
- Allergic shiners, sinusitis
- Mouth breathing, mouth sores, thrush
- **Dental**: delayed loss of baby teeth (hyper IgE)
- **Tonsils**: present/absent

Respiratory:
- Signs: hypoxia, tachypnea, clubbing

Cardiovascular:
- Congenital heart defects
- Murmur

Abdomen:
- Hepatosplenomegaly
- Perianal disease

Adenopathy:
- **Lymph nodes**: present/enlarged

Dermatology:
- **Fungal infections**: nails, skin, mouth
- **Rashes**: erythroderma, Omenn syndrome, severe eczema

Musculoskeletal:
- Arthritis, rashes

	PHAGOCYTIC	COMPLEMENT	HUMORAL	COMBINED
PRESENTATION	**Recurrent abscesses** **Gram neg** pneumonia Early onset **IBD** Delayed **umbilical cord separation** **Mucositis**	**Early** – SLE, nephrotic syndrome **Late** – *N. meningitidis* **Encapsulated bacteria**	Recurrent **sino-otopulmonary** infections **Ab-mediated** autoimmunity **> 18** months of age	**Candidiasis** **Failure to thrive** **PJP** pneumonia Chronic **diarrhea** **3 months** of age **OPPORTUNISTIC** infections Abnormal NMS
EX.	Chronic granulomatous disease (GCD) Leukocyte adhesion deficiency (LAD)	C2 deficiency Properdin deficiency	X-linked agamma-globulinemia CVID	Severe combined immunodeficiency (SCID)
INVESTIGATIONS	CBCd **Neutrophil Oxidative Burst Index** (NOBI)	**CH50** (assesses hemolytic activity of MAC*) AH50 *membrane attack complex	**Lymphocyte subsets** (B-cell quantification) **IgGAME**	**Lymphocyte subsets** **TREC* level** **Mitogen stimulation assay** *T-cell receptor excision circles
TREATMENT	TMP-SMX Itraconazole (CGD) Bone marrow transplant (BMT)	Amoxicillin Vaccination – *Pneumococcal* & *Meningococcal*	Prophylactic antibiotics IVIG/SCIG	**Urgent referral to immunology** BMT, IVIG, TMP-SMX **ISOLATION**

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