



Domain Expert Annotation Manual

Annotation page can be accessed at: <https://d371-34-212-40-25.ngrok-free.app/>

General Guidelines

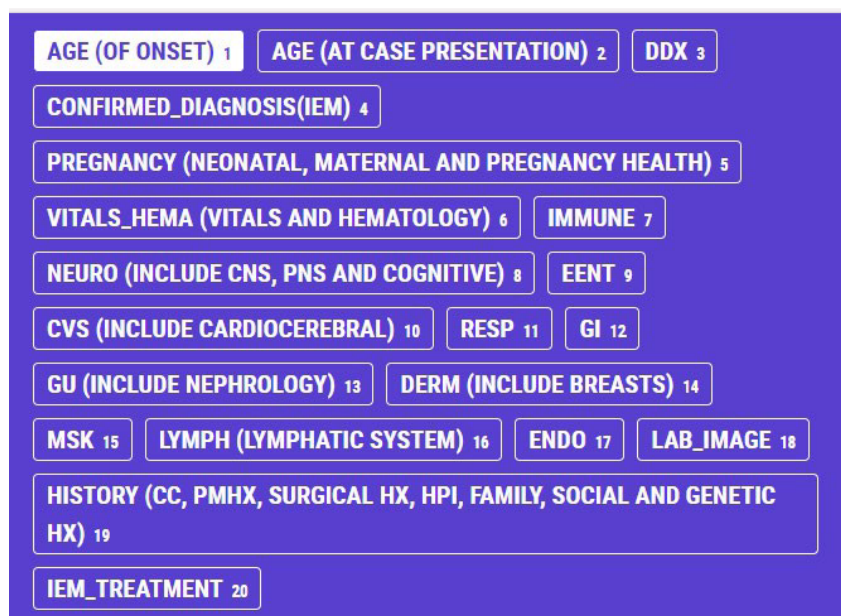
- **Please Include treatment to IEM only:**
Include Treatment details if they are directly relevant to the IEM diagnosis.
- **Please select complete phrases:** When annotating, capture entire phrases or sentences. Avoid splitting entities into separate parts; for example, if "elevated blood pressure of 140/90 mmHg" is mentioned, select the full phrase rather than isolating "blood pressure" and "140/90 mmHg."
- **Flexible labeling:** Information can fit into multiple categories or none at all. If a piece of information is relevant to more than one label, apply all applicable labels. If it does not fit any label, you may leave it unannotated.

 [Case Report]: "A 23 <u>year-old</u> female patient, 160 cm tall and 48 kg, was diagnosed with acute appendicitis and admitted for a	 [Case Report]: "A 23 <u>year-old</u> female patient, 160 cm tall and 48 kg, was diagnosed with acute appendicitis and admitted for a
---	---

Selecting Labels and Highlighting Text:

- **Select each label** that describes a span of text and highlight the corresponding words. There are **20 categories** available (see Figure 1).

Figure 1



Selecting and Deselecting Labels:

- To apply a label, click on the desired label below the highlighted phrase or sentence.
- To **deselect**, simply click on the label again.

Labeling Guidelines:

- You can apply one label at a time, but a piece of text can match **none, one, or multiple labels**
- For each labeled snippet, click the green “**tick**” mark at the bottom to confirm and move to the next snippet (see Figure 2).
- If you would like to revisit the case snippet at a later time, click on pause to skip to the next case first

Reviewing Completed Work:

- Your completed work appears in the History (Figure 3) bar. You can review up to **20 previous snippets**.

Saving Your Work:

- Click the **Save** icon (see Figure 4) to save your progress. **Important:** Once saved, all history will be cleared, and you won't be able to revisit previously completed snippets.

Figure 2:

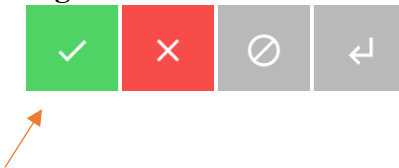


Figure 3:

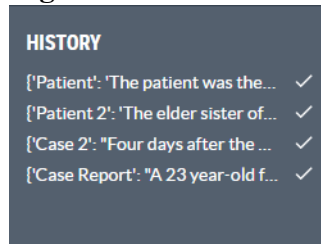


Figure 4:



Category Descriptions

1. **Pregnancy-Related Information (Pregnancy):**
 1. **Neonatal Health:** Record observations, conditions, or diagnostic findings specific to the infant's health.
 2. **Maternal Health:** Document the mother's health observations during pregnancy, excluding treatment details.
2. **Vital Signs and Hematology (Vitals_Hema):**
 1. **Vital Signs:** Include precise measurements of temperature, pulse, respiratory rate, blood pressure, and oxygen saturation (SpO2), along with units.
 2. **Hematology:** Document hematological conditions (e.g., anemia) and list blood test measurements such as hemoglobin, WBC count, and platelet count.
3. **Neurological and Cognitive Findings (Neuro):**
 1. **Neurological Findings:** Record specific observations or diagnoses related to neurological function.
 2. **Cognitive Findings:** Document any cognitive impairments or assessments.
4. **Eyes, Ears, Nose, and Throat (EENT):**
 1. **Eyes, Ears, Nose, Throat Findings:** Include observations or symptoms related specifically to each structure.
5. **Cardiovascular and Cerebrovascular System (CVS):**
 1. **Cardiac Findings:** List observations, signs, or symptoms directly related to the heart and cardiac function.
 2. **Vascular Findings:** Document vascular observations, including blood vessel-related conditions.
6. **Respiratory System (RESP):**
 1. **Respiratory Findings:** Record observations or symptoms directly related to respiratory function.
7. **Gastrointestinal System (GI):**
 1. **Gastrointestinal Findings:** Document observations, signs, or symptoms specific to the gastrointestinal system.
8. **Genitourinary System (GU):**
 1. **Urinary Findings:** Include observations specific to the urinary tract.
 2. **Genital Findings:** Document conditions specific to the genital organs.
9. **Dermatological Findings (DERM):**
 1. **Skin Conditions:** Include observations specific to skin conditions.
 2. **Facial Features:** Document any noteworthy observations related to facial features.
 3. **Breast Findings:** List any breast-related observations or conditions.
10. **Musculoskeletal System (MSK):**
 1. **Muscle Findings:** Record any observations or conditions related to muscle health.
 2. **Skeletal Findings:** Include observations specific to bones and joints.
11. **Lymphatic System (LYMPH):**

1. **Adenoid, Tonsils, and Lymphatic Tissues:** Record observations for each lymphatic structure individually.
 2. **Lymph Nodes, Thymus, Bone Marrow, Spleen:** Document findings specific to each structure.
 3. **Immune Cell Findings:** List immune cell-related observations or counts.
12. **Endocrine System (ENDO):**
1. **Endocrine Gland Findings:** Document any observations related to endocrine glands such as thyroid, adrenal glands, pancreas, etc.
13. **Laboratory and Imaging Results (Lab & Image)**
14. **Patient History (History):**
1. **Chief Complaint (CC):** Document the primary reason for seeking medical attention.
 2. **Past Medical History (PMHx):** Include known past medical conditions or diagnoses.
 3. **Past Surgical History:** List previous surgical procedures and outcomes.
 4. **History of Present Illness (HPI):** Provide a detailed narrative of the development of the current illness.
 5. **Family and Genetics History:** Include any family history of genetic conditions or hereditary diseases.
 6. **Social History:** Document lifestyle factors such as smoking, alcohol use, occupation, and living conditions.

References:

Span Categorization (<https://prodi.gy/docs/span-categorization>)

Interface Functions: (<https://prodi.gy/docs/api-web-app>)

Amendments for Reconciliation

Negative Findings

- Include negative findings (e.g., "normal development," "no dark urine") to ensure a comprehensive record of what has been assessed. This avoids ambiguity and supports diagnostic precision.

Dental Manifestations

- **EENT:** Categorize dental conditions involving the teeth, gums, or soft tissues of the oral cavity under EENT. Examples include dental caries, periodontal disease, oral infections, and abscesses.
- **MSK:** Use MSK for conditions affecting the jawbone, temporomandibular joint (TMJ), or surrounding musculoskeletal structures. Examples include jaw fractures, TMJ disorders, or structural abnormalities of the jaw.
- **Recommendation:** Prioritize categorizing dental conditions under EENT unless explicitly related to the skeletal framework (e.g., jaw or TMJ).

History Section

- **Prioritize PMH and Surgical History:** Ensure exhaustive listing of past medical and surgical histories, as they provide critical diagnostic information. Include all relevant conditions and surgeries verbatim.
- **HPI and Chief Complaint:** Focus on concise summaries of the chief complaint, its onset, progression, and associated symptoms. Avoid excessive narratives unless they provide significant diagnostic value.
- **Family and Social History:** Keep these sections concise, focusing on elements directly relevant to the diagnosis.

GU vs. Endocrine System

- **Endocrine (ENDO):**
 - Use for endocrine gland dysfunctions or hormonal disorders.
 - Include systemic manifestations originating from endocrine dysfunction affecting genital organs (e.g., congenital adrenal hyperplasia, hypogonadism).
 - Lab tests or imaging related to endocrine issues should remain categorized under "Lab_Image."
- **GU:**
 - Use for structural, infectious, or non-hormonal genital abnormalities (e.g., UTI, structural genital conditions).
 - Contexts related to menstruation should be categorized based on the underlying cause:
 - Hormonal cause → **ENDO**.
 - Structural or infectious cause → **GU**.

Musculoskeletal (MSK) Observations

- **Muscle Weakness:** Categorize muscle strength observations under NEURO by default unless explicitly attributed to muscle-specific conditions (e.g., injury, myopathy).

ECG Placement

- Include ECG findings under the **Lab_Image** category. While ECG is not a conventional imaging modality like MRI or CT, its diagnostic data aligns with other lab-based results.