

SUPPLEMENTARY APPENDIX

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Table S1: Disease categories of the NEJM dataset

Disease Classification	Number
Infectious Diseases	36
Autoimmune Diseases	20
Neoplastic Diseases (Tumors)	19
Genetic Diseases, Congenital Diseases	19
Iatrogenic Diseases	15
Systemic Diseases	11
Nervous System Diseases	9
Digestive System Diseases	7
Endocrine and Metabolic Diseases	6
Blood System Diseases	6
Mental Illness or Psychological Disorders and Related Trauma	6
Cardiovascular System Diseases	2
Respiratory System Diseases	2
Trauma-related Diseases	2
Poisoning, Addictive Substance, or Drug Abuse	2
Malnutrition	2

Table S2. Diagnosis classification criteria of the offline dataset

ID	Disease Type	Diagnosis Classification Criteria		
		Class 1.1	Class 1.2	Class 2
1	Systemic scleroderma	Systemic sclerosis/Scleroderma/ Scleroderma with gastroesophageal involvement/ Systemic sclerosis (Limited cutaneous type CREST Syndrome)	-	-
2	Ménétrier's disease	Menetrier's disease		Hypertrophic gastropathy/ Hypertrophic gastritis
3	Obscure source of bleeding: Pancreatic duct stones causing bleeding	Pancreatic duct stones	-	Chronic pancreatitis/ Chronic Pancreatitis Complications/ Hemosuccus pancreaticus
4	Congenital esophageal stenosis	Congenital esophageal stenosis		
5	Autoimmune enteropathy	Autoimmune enteropathy		
6	Very early-onset inflammatory bowel disease (IL-10RA gene deficiency)	Very early onset Crohn's disease/ IL-10 Receptor Deficiency		
7	Cronkhite-Canada syndrome	Cronkhite-Canada syndrome (CCS)		
8	Hepatic myelopathy	Hepatic myelopathy/Portosystemic Myelopathy		
9	Abdominal cocoon syndrome	Abdominal cocoon/ sclerosing encapsulating peritonitis/ Encapsulating Peritoneal Sclerosis		
10	Jackhammer esophagus	Jackhammer esophagus	Hypercontractile esophagus	
11	Cronkhite-Canada syndrome	Cronkhite-Canada syndrome		

12	Glycogen storage disease	Glycogen storage disease/ Von Gierke disease		
13	Cowden syndrome	Cowden syndrome/Multiple Hamartoma syndrome		
14	Mycophenolate mofetil-associated colitis	Mycophenolate mofetil-associated colitis	Drug-induced colitis	
15	Idiopathic mesenteric phleboscrotic colitis	Idiopathic mesenteric phleboscrotic colitis /Idiopathic Mesenteric Phleboscrosis	Phleboscrotic Colitis	
16	Small bowel diaphragm disease	Small bowel diaphragm disease/NSAID-induced Small Intestinal Diaphragm Disease/ NSAID-induced diaphragm disease		Nonsteroidal anti-inflammatory drug (NSAID) enteropathy/ NSAID-induced enteropathy leading to intestinal strictures
17	Gardner syndrome	Gardner syndrome		Familial adenomatous polyposis (FAP)
18	Diffuse esophageal spasm	Diffuse esophageal spasm		
19	Ménétrier's disease	Menetrier's disease		
20	Abdominal cocoon syndrome	Abdominal cocoon/ sclerosing encapsulating peritonitis/ Encapsulating peritoneal sclerosis		
21	Refractory diarrhea etiology: Clostridium difficile infection	Clostridium difficile infection/	Pseudomembranous colitis	Medication-induced diarrhea/ Antibiotic-associated colitis,
22	Lymphocytic esophagitis	Lymphocytic esophagitis		
23	Hepatoid adenocarcinoma of the stomach	Hepatoid adenocarcinoma of the stomach /Gastric cancer (hepatoid adenocarcinoma subtype)		AFP-producing gastric carcinoma
24	Gastric tuberculosis	Gastric tuberculosis	Reactivation of pulmonary tuberculosis	
25	Gardner syndrome	Gardner syndrome		Familial adenomatous

				polyposis (FAP)
26	Acute intermittent porphyria	Acute intermittent porphyria	Porphyria	
27	Citrullinemia (Urea cycle disorder)	Urea cycle disorder/	Late-onset ornithine transcarbamylase (OTC) Deficiency	
28	Abdominal pain etiology: Porphyria	Porphyria		
29	Mitochondrial neurogastrointestinal encephalomyopathy	Mitochondrial neurogastrointestinal encephalomyopathy	Mitochondrial Myopathy/ Mitochondrial Disease	
30	Hereditary angioedema	Hereditary angioedema		
31	Left ventricular non-compaction cardiomyopathy	Left ventricular non-compaction cardiomyopathy	Myocardial Non-compaction Cardiomyopathy	
32	Primary porphyria	Porphyria		
33	Hereditary hemorrhagic telangiectasia	Hereditary hemorrhagic telangiectasia/ Osler-Weber-Rendu syndrome		
34	Primary myelofibrosis	Primary Myelofibrosis	Myelofibrosis	
35	Multiple myeloma	Multiple myeloma/ Plasma Cell Myeloma		
36	Primary non-Hodgkin lymphoma of the liver (Diffuse, T-cell phenotype)	Primary non-Hodgkin lymphoma of the liver	Lymphoma	
37	Extranodal NK/T-cell lymphoma	Extranodal NK/T-cell lymphoma	Intestinal T-cell lymphoma/ Enteropathy-associated T-cell lymphoma/ lymphoma	
38	Systemic mastocytosis (Involving the gastrointestinal tract)	Systemic mastocytosis/ Mastocytosis		

39	Erythropoietic protoporphyria	Erythropoietic protoporphyria	Protoporphyria	
40	Waldenström macroglobulinemia	Waldenström macroglobulinemia/ Lymphoplasmacytic Lymphoma		Lymphoma
41	Multisystem Langerhans cell histiocytosis	Multisystem Langerhans cell histiocytosis /Langerhans cell histiocytosis		
42	Small intestinal T-cell lymphoma	Small intestinal T-cell lymphoma	Lymphoma	
43	Henoch-Schönlein purpura (Abdominal type)	Henoch-Schönlein Purpura		
44	Antiphospholipid syndrome	Antiphospholipid syndrome		
45	Felty's syndrome (Rheumatoid arthritis-splenomegaly syndrome)	Felty's syndrome/ Rheumatoid arthritis-splenomegaly syndrome		Rheumatoid arthritis/ Rheumatoid arthritis with secondary splenomegaly
46	Tacrolimus-induced vasculitis/Systemic vasculitis	Tacrolimus-induced vasculitis/Systemic vasculitis	Adverse reaction to tacrolimus and Vasculitis	Vasculitis/ Medication-induced Side Effects (Tacrolimus-related)
47	Systemic lupus erythematosus	Systemic lupus erythematosus		
48	Selective IgA deficiency (with Giardia infection)	Selective IgA deficiency (with Giardia infection)	Selective IgA deficiency	Common Variable Immunodeficiency
49	IgG4-related disease	IgG4-related disease		
50	Congenital immunodeficiency disorders	Congenital immunodeficiency disorders		Common Variable Immunodeficiency
51	Eosinophilic granulomatosis with polyangiitis	Eosinophilic granulomatosis with polyangiitis/ Churg-Strauss Syndrome		Granulomatosis with polyangiitis/Microscopic polyangiitis
52	Henoch-Schönlein	Henoch-Schönlein		

	purpura	purpura		
53	POEMS syndrome	POEMS syndrome		
54	Systemic sclerosis (CREST syndrome subtype)	Systemic sclerosis (CREST syndrome subtype)	Systemic sclerosis/ Scleroderma	
55	Paraneoplastic syndrome (Trousseau syndrome associated with metastatic cancer of unknown origin)	Paraneoplastic syndrome (Trousseau syndrome associated with metastatic cancer of unknown origin)/ Trousseau syndrome		Cancer with paraneoplastic syndrome leading to DIC and multiorgan dysfunction/ Metastatic cancer with paraneoplastic syndrome /Metastatic Carcinoma with Associated Thromboembolism
56	Systemic primary amyloidosis	Systemic primary amyloidosis	Amyloidosis	
57	Primary amyloidosis (AL type)	Primary amyloidosis (AL type)	Amyloidosis	
58	Systemic sclerosis	Systemic sclerosis/ Scleroderma		
59	Light chain deposition disease of the small intestine	Light chain deposition disease/ κ Light chain deposition disease		
60	Systemic light chain amyloidosis	Systemic light chain amyloidosis	Amyloidosis	
61	POEMS	POEMS		
62	Renal tubular acidosis (Distal renal tubular acidosis)	Renal tubular acidosis (Distal renal tubular acidosis)/ Distal Renal Tubular Acidosis (Type I RTA)	Type IV renal tubular acidosis (RTA)	
63	Hemophagocytic Lymphohistiocytosis (Evolving from Crohn's disease)	Hemophagocytic Lymphohistiocytosis (Evolving from Crohn's disease)/ Hemophagocytic Lymphohistiocytosis and Crohn's disease	Hemophagocytic Lymphohistiocytosis (HLH)	
64	Type I renal tubular acidosis (Distal renal tubular acidosis)	Type I renal tubular acidosis (Distal renal tubular acidosis)		

65	Hemophagocytic syndrome (Evolving from colonic ulcers)	Hemophagocytic syndrome (Evolving from colonic ulcers)/	Hemophagocytic Lymphohistiocytosis (HLH)	
66	Lead poisoning (Causing recurrent pseudo-intestinal obstruction and anemia)	Lead poisoning		
67	Hemorrhagic fever with renal syndrome	Hemorrhagic fever with renal syndrome	Hantavirus infection/ Hantavirus pulmonary syndrome	

Table S3: Performance of different LLMs using different prompts in the NEJM dataset

LLMs	Prompt	Average Number of Possible Diagnoses (SD*)	Accuracy (%, 95% CI*)	Coverage (%, 95% CI*)
Claude 3.5 Sonnet	Answer Only	5.1 (0.4)	24.5 (20.9-28.5)	55.1 (50.7-59.4)
	A&R	5.0 (0.0)	32.2 (28.2-36.4)	67.8 (63.6-71.8)
	CoT	5.0 (0.0)	35.7 (31.6-40.0)	67.5 (63.2-71.4)
Claude 3 Opus	Answer Only	5.1 (0.3)	22.0 (18.6-25.8)	53.1 (48.8-57.5)
	A&R	5.0 (0.1)	28.8 (25.0-32.9)	59.6 (55.3-63.8)
	CoT	5.0 (0.0)	31.0 (27.1-35.2)	62.7 (58.4-66.9)
GPT-4o	Answer Only	5.0 (0.0)	13.9 (11.2-17.2)	37.5 (33.3-41.8)
	A&R	5.0 (0.1)	21.8 (18.4-25.6)	48.2 (43.9-52.6)
	CoT	5.0 (0.2)	17.1 (14.0-20.6)	42.0 (37.7-46.3)
GPT-3.5t	Answer Only	5.0 (0.1)	4.1 (2.7-6.2)	17.5 (14.4-21.0)
	A&R	4.8 (0.5)	7.1 (5.1-9.6)	30.4 (26.5-34.5)
	CoT	4.4 (1.0)	9.2 (7.0-12.1)	29.6 (25.8-33.7)
Gemini-1.5-Pro	Answer Only	5.2 (1.2)	14.9 (12.1-18.3)	40.8 (36.6-45.1)
	A&R	4.5 (0.7)	18.8 (15.6-22.5)	50.8 (46.4-55.1)
	CoT	5.0 (0.3)	18.6 (15.5-22.3)	51.2 (46.8-55.5)

*SD: standard deviation

Table S4: Performance of different LLMs using different prompts in the offline dataset

LLMs	Prompt	Average Number of Possible Diagnoses (SD*)	Coverage (% 95% CI*)	Accuracy (% 95% CI*)
Claude 3.5 Sonnet	Answer Only	4.9 (0.9)	75.1 (68.6-80.7)	51.2 (44.2-58.2)
	A&R	5.0 (0.0)	79.6 (73.4-84.7)	53.2 (46.2-60.1)
	CoT	5.0 (0.0)	81.1 (75.0-86.0)	53.2 (46.2-60.1)
Claude 3 Opus	Answer Only	5.0 (0.5)	68.2 (61.3-74.3)	47.3 (40.3-54.3)
	A&R	5.0 (0.0)	73.6 (67.0-79.4)	49.3 (42.3-56.2)
	CoT	5.0 (0.0)	67.2 (60.3-73.4)	42.3 (35.5-49.3)
GPT-4o	Answer Only	5.0 (0.5)	43.8 (37.0-50.8)	23.4 (18.0-29.8)
	A&R	5.0 (0.0)	48.8 (41.8-55.8)	26.4 (20.6-33.0)
	CoT	5.0 (0.0)	53.2 (46.2-60.1)	29.4 (23.4-36.1)
GPT-3.5t	Answer Only	4.9 (0.5)	29.4 (23.4-36.1)	11.4 (7.7-16.7)
	A&R	4.9 (0.3)	30.3 (24.3-37.1)	10.9 (7.3-16.1)
	CoT	4.6 (0.8)	34.8 (28.5-41.8)	15.4 (11.0-21.2)
Gemini-1.5-Pro	Answer Only	5.0 (0.7)	51.2 (44.2-58.2)	27.9 (22.0-34.5)
	A&R	5.0 (0.3)	52.7 (45.7-59.7)	25.4 (19.7-31.9)
	CoT	5.1 (0.2)	48.3 (41.3-55.3)	27.9 (22.0-34.5)

*SD: standard deviation

Table S5: Performance of Sonnet Pyramid Framework in the NEJM dataset

	Accuracy (% , 95% CI)	Coverage (% , 95% CI)
Multiple LLMs		
A&R Chain		
Layer 1	21.4 (18.0-25.2)	57.5 (53.1-61.7)
Layer 2	31.6 (27.7-35.8)	67.3 (63.0-71.2)
Layer 3	32.4 (28.4-36.6)	67.6 (63.4-71.6)
A&R Tree		
Layer 1	34.9 (30.9-39.2)	69.6 (65.5-73.5)
Layer 2	35.3 (31.2-39.6)	70.6 (66.5-74.4)
Layer 3	35.3 (31.2-39.6)	70.6 (66.5-74.4)
CoT Tree		
Layer 1	36.7 (32.6-41.0)	69.0 (64.8-72.9)
Layer 2	36.9 (32.8-41.2)	68.2 (64.0-72.2)
Layer 3	37.5 (33.3-41.8)	69.6 (65.5-73.5)
Merge		
Layer 1	40.0 (35.8-44.3)	72.2 (68.1-75.9)
Layer 2	40.4 (36.2-44.7)	74.5 (70.5-78.1)
Layer 3	44.1 (39.8-48.5)	77.6 (73.8-81.1)
Sonnet		
A&R Chain		
Layer 1	36.9 (32.8-41.2)	69.2 (65.0-73.1)
Layer 2	38.2 (34.1-42.6)	70.6 (66.5-74.4)
Layer 3	38.2 (34.1-42.6)	70.6 (66.5-74.4)
A&R Tree		
Layer 1	35.9 (31.8-40.2)	69.4 (65.2-73.3)
Layer 2	37.6 (33.5-42.0)	71.8 (67.7-75.5)
Layer 3	37.5 (33.3-41.8)	71.8 (67.7-75.5)
CoT Tree		
Layer 1	36.7 (32.6-41.0)	68.2 (64.0-72.2)
Layer 2	37.3 (33.1-41.6)	68.8 (64.6-72.7)
Layer 3	37.6 (33.5-42.0)	69.8 (65.7-73.7)
Merge		
Layer 1	41.2 (37.0-45.5)	72.2 (68.1-75.9)
Layer 2	42.2 (37.9-46.5)	73.5 (69.5-77.2)
Layer 3	44.7 (40.4-49.1)	75.5 (71.5-79.1)
Output		
Output	46.1 (41.8-50.5)	79.0 (75.2-82.4)

Table S6: Performance of Sonnet Pyramid Framework in the offline dataset

	Accuracy (% , 95% CI)	Coverage (% , 95% CI)
Multiple LLMs		
A&R Chain		
Layer 1	34.3 (28.0-41.3)	58.2 (51.2-64.9)
Layer 2	50.7 (43.8-57.7)	70.6 (63.9-76.6)
Layer 3	50.7 (43.8-57.7)	74.1 (67.5-79.8)
A&R Tree		
Layer 1	53.7 (46.7-60.6)	78.1 (71.8-83.4)
Layer 2	54.7 (47.7-61.6)	82.1 (76.1-86.9)
Layer 3	58.7 (51.7-65.4)	84.6 (78.8-89.0)
CoT Tree		
Layer 1	56.2 (49.2-63.0)	80.6 (74.5-85.6)
Layer 2	56.2 (49.2-63.0)	81.6 (75.6-86.4)
Layer 3	58.7 (51.7-65.4)	82.6 (76.6-87.3)
Merge		
Layer 1	62.7 (55.7-69.2)	84.1 (78.3-88.6)
Layer 2	61.7 (54.7-68.3)	86.6 (81.1-90.7)
Layer 3	64.7 (57.7-71.1)	89.6 (84.5-93.1)
Sonnet		
A&R Chain		
Layer 1	59.2 (52.2-65.9)	82.1 (76.1-86.9)
Layer 2	56.7 (49.7-63.5)	83.1 (77.2-87.7)
Layer 3	57.2 (50.2-64.0)	83.6 (77.7-88.2)
A&R Tree		
Layer 1	55.7 (48.7-62.5)	80.1 (73.9-85.1)
Layer 2	56.7 (49.7-63.5)	84.1 (78.3-88.6)
Layer 3	56.2 (49.2-63.0)	84.1 (78.3-88.6)
CoT Tree		
Layer 1	58.7 (51.7-65.4)	81.6 (75.6-86.4)
Layer 2	59.2 (52.2-65.9)	83.6 (77.7-88.2)
Layer 3	61.2 (54.2-67.8)	85.6 (80.0-89.9)
Merge		
Layer 1	61.7 (54.7-68.3)	86.1 (80.5-90.3)
Layer 2	61.2 (54.2-67.8)	88.1 (82.8-91.9)
Layer 3	61.7 (54.7-68.3)	89.6 (84.5-93.1)
Output		
Output	63.7 (56.7-70.1)	91.0 (86.2-94.3)

Table S7. Diagnostic performance of o1-mini and o3-mini using the Pyramid Framework, direct prompting, and CoT in the NEJM dataset

NEJM Dataset	Average Number of Possible Diagnoses (SD)	Accuracy (% CI)	95% Coverage (% CI)
o1-mini			
Direct prompting	5.4 (0.8)	26.9 (23.2-30.9)	53.9 (49.5-58.2)
CoT	5.0 (0.4)	22.7 (19.3-26.6)	51.8 (47.4-56.1)
Pyramid Framework	5.1 (0.5)	32.4 (28.4-36.6)	65.9 (61.6-69.9)
o3-mini			
Direct prompting	5.0 (0.5)	21.8 (18.4-25.6)	46.1 (41.8-50.5)
CoT	5.0 (0.1)	19.6 (16.4-23.3)	42.9 (38.7-47.3)
Pyramid Framework	5.0 (0)	39.4 (35.2-43.7)	70.6 (66.5-74.4)

Table S8. Diagnostic performance of o1-mini and o3-mini using the Pyramid Framework, direct prompting, and CoT in the offline dataset

Offline Dataset	Average Number of Possible Diagnoses (SD)	Accuracy (% , 95% CI)	Coverage (% , 95% CI)
o1-mini			
Direct prompting	5.3 (0.7)	50.2 (43.3-57.2)	77.6 (71.3-82.9)
CoT	5.0 (0.1)	42.8 (36.0-49.8)	69.2 (62.3-75.3)
Pyramid Framework	5.1 (0.4)	62.2 (55.2-68.7)	88.6 (83.3-92.3)
o3-mini			
Direct prompting	5.1 (1.0)	40.8 (34.1-47.8)	57.7 (50.7-64.5)
CoT	5.0 (0.1)	35.8 (29.4-42.8)	51.2 (44.2-58.2)
Pyramid Framework	5.0 (0)	65.7 (58.7-72.0)	86.6 (81.1-90.7)

Table S9: Average response token count of the Sonnet Pyramid Framework

	NEJM Dataset	Offline Dataset
	Average token count *	Average token count
Single Round		
Sonnet A&R	276	322
Sonnet CoT	761	821
Modules		
Multiple		
A&R Chain	1379	1349
A&R Tree	3638	4159
CoT Tree	5912	6185
Sonnet		
A&R Chain	1308	1325
A&R Tree	3420	3770
CoT Tree	5456	5746
Merge Layer		
Merge Layer 2	11731	12477
Merge Layer 3	35794	38090
Output	74394	79405

*: Token count was calculated using BPE algorithm.

Table S10: Performance of Sonnet Pyramid Framework in the JAMA dataset

JAMA Dataset	Accuracy (% , 95% CI)	
	Zero-shot CoT	Pyramid
Total	75.9 (74.7-77.2)	74.1 (72.8-75.4)
Round 1	76.0 (73.7-78.1)	73.7 (71.4-75.9)
Round 2	76.1 (73.8-78.2)	74.0 (71.7-76.2)
Round 3	75.7 (73.5-77.9)	74.5 (72.2-76.7)

Table S11: International standard book numbers of the chosen medical case books*

Number of Medical Case Books	International Standard Book Number
1	978-7-117-13071-4
2	978-7-5189-4773-7
3	978-7-117-16571-6
4	978-7-5679-1513-8
5	978-7-81072-882-9
6	978-7-03-061629-6
7	978-7-5439-8460-8
8	978-7-5641-9312-6
9	978-7-5377-2957-4
10	978-7-8113-6080-6
11	978-7-2290-4120-5

*All chosen case books were published by national top-tier tertiary hospitals.

Table S12: LLMs information and query date

Model Type	Specific Model Number	Temperature	Query Date
GPT-3.5t	gpt-3.5-turbo-0613	1.0	September, 2024
GPT-4o	gpt-4o-2024-08-06	1.0	September, 2024
o1-mini	o1-mini-2024-09-12	1.0	December, 2024
o3-mini	o3-mini-medium	1.0	February, 2025
Gemini-1.5-Pro	gemini-1.5-pro-latest	1.0	September, 2024
Claude 3 Opus	claude-3-opus-20240229	1.0	September, 2024
Claude 3.5 Sonnet	claude-3-5-sonnet-20240620	1.0	September, 2024

Table S13. Diagnosis classification criteria of NEJM dataset

DOI	Diagnosis	Diagnosis Classification Criteria		
		Class 1.1	Class 1.2	Class 2
10.1056/NEJMcpc1900142	Hyperthyroidism due to Graves' disease.	Hyperthyroidism due to Graves' disease.	Thyroid Storm/Hyperthyroidism	
10.1056/NEJMcpc1900419	Systemic lupus erythematosus.	Systemic lupus erythematosus.		
10.1056/NEJMcpc1900420	Intraparenchymal and intracranial hemorrhage presumably due to birth trauma.	Intraparenchymal and intracranial hemorrhage presumably due to birth trauma/Birth-related subdural hemorrhage with possible recent exacerbation		
10.1056/NEJMcpc1900421	Metastatic postpubertal immature teratoma.	Metastatic postpubertal immature teratoma.		Soft tissue sarcoma
10.1056/NEJMcpc1900423	Active myocarditis consistent with myocarditis related to immune checkpoint inhibition.	Active myocarditis consistent with myocarditis related to immune checkpoint inhibition.		
10.1056/NEJMcpc1900589	Statin-associated autoimmune myopathy.	Statin-associated autoimmune myopathy.		
10.1056/NEJMcpc1900590	Concussion, benign paroxysmal positional vertigo, and attention deficit–hyperactivity disorder.	Concussion, benign paroxysmal positional vertigo, and attention deficit–hyperactivity disorder.		Post-Concussion Syndrome
10.1056/NEJMcpc1900591	Surreptitious ingestion of isopropyl alcohol	Surreptitious ingestion of isopropyl alcohol		
10.1056/NEJMcpc1900594	Intestinal tuberculosis	Intestinal tuberculosis	Tuberculosis	
10.1056/NEJMcpc1900595	Tropical pulmonary	Tropical pulmonary		

	eosinophilia	eosinophilia		
10.1056/NEJMcpc1900596	Acute suppurative appendicitis and periappendicitis.	Acute suppurative appendicitis and periappendicitis.	Acute appendicitis	
10.1056/NEJMcpc1900597	Leber's hereditary optic neuropathy	Leber's hereditary optic neuropathy		
10.1056/NEJMcpc1900598	Flail mitral valve due to acute myocardial infarction of the papillary muscle in the absence of obstructive coronary artery disease	Flail mitral valve due to acute myocardial infarction of the papillary muscle in the absence of obstructive coronary artery disease		Acute myocardial infarction
10.1056/NEJMcpc1904039	Disseminated cryptococcosis.	Disseminated cryptococcosis.		
10.1056/NEJMcpc1904040	Chronic Chagas' disease with reactivation of latent Trypanosoma cruzi infection	Chronic Chagas' disease with reactivation of latent Trypanosoma cruzi infection		
10.1056/NEJMcpc1904041	Interstitial lung disease associated with FLNA mutation	Interstitial lung disease associated with FLNA mutation/Filamin A (FLNA) Gene-Related Lung Disease		
10.1056/NEJMcpc1904042	Lymphocytic choriomeningitis virus infection	Lymphocytic choriomeningitis virus infection		
10.1056/NEJMcpc1904043	Opioid use disorder and malingering.	Opioid use disorder and malingering.		Opioid use disorder and factitious disorder.
10.1056/NEJMcpc1904045	Adenovirus (serotype 2) meningoencephalitis	Adenovirus (serotype 2) meningoencephalitis		Viral meningoencephalitis
10.1056/NEJMcpc1904046	Amniotic fluid embolism	Amniotic fluid embolism		
10.1056/NEJMcpc1904047	B-cell acute lymphoblastic leukemia.	B-cell acute lymphoblastic leukemia.		
10.1056/NEJMcpc1904048	Juvenile myelomonocytic leukemia	Juvenile myelomonocytic leukemia		leukemia

10.1056/NEJMcp1904049	Congenital esophageal stenosis with fibromuscular thickening of the esophagus	Congenital esophageal stenosis with fibromuscular thickening of the esophagus	Congenital esophageal stenosis	
10.1056/NEJMcp1909620	Undifferentiated autoimmune rheumatic disease.	Undifferentiated autoimmune rheumatic disease.		Systemic Lupus Erythematosus
10.1056/NEJMcp1909621	Aspergillus terreus infection associated with toxic effects induced by immune checkpoint inhibitor therapy (pneumonitis, colitis, and motor axonal neuropathy) and metastatic melanoma without evidence of active disease	Aspergillus terreus infection associated with toxic effects induced by immune checkpoint inhibitor therapy (pneumonitis, colitis, and motor axonal neuropathy) and metastatic melanoma without evidence of active disease		Immune Checkpoint Inhibitor-Related Pneumonitis (ICI-Pneumonitis)/
10.1056/NEJMcp1909623	Proteinase 3 antineutrophil cytoplasmic antibody–associated vasculitis consistent with granulomatosis with polyangiitis.	Proteinase 3 antineutrophil cytoplasmic antibody–associated vasculitis consistent with granulomatosis with polyangiitis/Granulomatosis with polyangiitis		
10.1056/NEJMcp1909624	Creutzfeldt–Jakob disease	Creutzfeldt–Jakob disease		
10.1056/NEJMcp1909625	Kaposi’s sarcoma of the gastrointestinal tract.	Kaposi’s sarcoma of the gastrointestinal tract.		
10.1056/NEJMcp1909627	Skin involvement associated with peripheral T-cell lymphoma most consistent with primary cutaneous gamma–delta T-cell lymphoma	Skin involvement associated with peripheral T-cell lymphoma most consistent with primary cutaneous gamma–delta T-cell lymphoma	Lymphoma	
10.1056/NEJMcp1909628	antiphospholipid antibody syndrome; Diffuse	Antiphospholipid antibody syndrome; Diffuse		

	pulmonary alveolar hemorrhage with focal infarction, multifocal thromboembolic disease, and neutrophilic infiltrate.	pulmonary alveolar hemorrhage with focal infarction, multifocal thromboembolic disease, and neutrophilic infiltrate.		
10.1056/NEJMcpc1913468	Disseminated coccidioidomycosis and ANCA-associated glomerulonephritis.	Disseminated coccidioidomycosis and ANCA-associated glomerulonephritis.		Disseminated coccidioidomycosis/ANCA-associated glomerulonephritis/Microscopic Polyangiitis (MPA)/Granulomatosis with Polyangiitis
10.1056/NEJMcpc1913469	Autoimmune primary adrenal insufficiency and celiac disease	Autoimmune primary adrenal insufficiency and celiac disease		Autoimmune primary adrenal insufficiency/Primary Adrenal Insufficiency (Addison's Disease)/Celiac disease
10.1056/NEJMcpc1913471	Antiepileptic drug toxicity	Antiepileptic drug toxicity/Antiepileptic Drug-Induced QRS Widening	Sodium Channel Blocker Toxicity (Lacosamide, Oxcarbazepine)	Drug-induced wide-complex tachycardia
10.1056/NEJMcpc1913472	Human immunodeficiency virus type 2 infection and cerebral toxoplasmosis	Human immunodeficiency virus type 2 infection and cerebral toxoplasmosis		Toxoplasmic Encephalitis/HIV
10.1056/NEJMcpc1913473	Strongyloidiasis with human T-lymphotropic virus type 1 infection.	Strongyloidiasis with human T-lymphotropic virus type 1 infection.		Strongyloidiasis
10.1056/NEJMcpc1913474	Disseminated Mycobacterium bovis infection	Disseminated Mycobacterium bovis infection		Tuberculosis
10.1056/NEJMcpc1913475	Maturity-onset diabetes of the young due to a GCK variant	Maturity-onset diabetes of the young due to a GCK variant/Maturity-Onset		

		Diabetes of the Young		
10.1056/NEJMcp1913476	Ulcerative colitis.	Ulcerative colitis.		Inflammatory bowel disease
10.1056/NEJMcp1913477	LGII autoimmune encephalitis.	LGII autoimmune encephalitis.	Autoimmune encephalitis	
10.1056/NEJMcp1916250	Anaplasma phagocytophilum infection.	Anaplasma phagocytophilum infection.		Tick-borne disease
10.1056/NEJMcp1916252	AA amyloidosis complicated by cerebral mucormycosis.	AA amyloidosis complicated by cerebral mucormycosis.		Cerebral Mucormycosis
10.1056/NEJMcp1916254	Crescentic IgA nephropathy	Crescentic IgA nephropathy		IgA Nephropathy (Berger's Disease)
10.1056/NEJMcp1916256	Hantavirus cardiopulmonary syndrome	Hantavirus cardiopulmonary syndrome	Hantavirus Infection	
10.1056/NEJMcp1916257	Pancreatitis, panniculitis, and polyarthritis syndrome.	Pancreatitis, panniculitis, and polyarthritis syndrome.		Pancreatitis and panniculitis
10.1056/NEJMcp1916258	Pulmonary histoplasmosis.	Pulmonary histoplasmosis/Histoplasmosis		
10.1056/NEJMcp2002412	Aceruloplasminemia, hereditary.	Aceruloplasminemia, hereditary.		
10.1056/NEJMcp2002413	Post-traumatic stress disorder.	Post-traumatic stress disorder.		
10.1056/NEJMcp2002415	IgG4-related disease, with tubulointerstitial nephritis, membranous nephropathy, and inflammatory pseudotumor involving the lacrimal gland	IgG4-related disease, with tubulointerstitial nephritis, membranous nephropathy, and inflammatory pseudotumor involving the lacrimal gland	IgG4-related disease	
10.1056/NEJMcp2002416	Systemic primary amyloidosis.	Systemic primary amyloidosis/Amyloidosis		
10.1056/NEJMcp2002420	Pseudohyperaldosteronism suggestive of excessive	Pseudohyperaldosteronism suggestive of excessive		Drug-Induced Arrhythmia

	licorice consumption complicated by cardiac arrest associated with ventricular fibrillation	licorice consumption complicated by cardiac arrest associated with ventricular fibrillation/Severe hypokalemia-induced cardiac arrest due to excessive licorice consumption/Toxic cardiomyopathy from licorice ingestion		
10.1056/NEJMcp2004979	Necrotizing granulomatous lymphadenitis due to Mycobacterium tuberculosis complex	Necrotizing granulomatous lymphadenitis due to Mycobacterium tuberculosis complex	Tuberculous lymphadenitis	Tuberculosis
10.1056/NEJMcp2004981	Post-traumatic stress disorder and major depressive disorder in the context of culturally and religiously derived beliefs and behaviors	Post-traumatic stress disorder and major depressive disorder in the context of culturally and religiously derived beliefs and behaviors/Post-traumatic stress disorder and major depressive disorder		Post-traumatic stress disorder
10.1056/NEJMcp2004991	Rasburicase-induced methemoglobinemia in the context of glucose-6-phosphate dehydrogenase deficiency. IgA lambda multiple myeloma.	Rasburicase-induced methemoglobinemia in the context of glucose-6-phosphate dehydrogenase deficiency. IgA lambda multiple myeloma.		Multiple myeloma
10.1056/NEJMcp2004992	Cerebellar ataxia, neuropathy, and vestibular areflexia syndrome due to a biallelic expansion in RFC1.	Cerebellar ataxia, neuropathy, and vestibular areflexia syndrome (CANVAS) due to a biallelic		

		expansion in RFC1		
10.1056/NEJMcp2004996	Borrelia miyamotoi infection.	Borrelia miyamotoi infection.		Lyme disease/Tick-borne disease
10.1056/NEJMcp2027077	Prosthetic paravalvular leak	Prosthetic paravalvular leak		Mechanical Valve–Associated Hemolytic Anemia
10.1056/NEJMcp2027078	Vitamin D deficiency	Vitamin D deficiency	Nutritional Rickets	
10.1056/NEJMcp2027080	Seizure from neurocysticercosis.	Seizure from neurocysticercosis/Neurocysticercosis		
10.1056/NEJMcp2027083	Cryptococcal meningoencephalitis and advanced human immunodeficiency virus infection.	Cryptococcal meningoencephalitis and advanced human immunodeficiency virus infection.		Cryptococcal meningoencephalitis/HIV infection
10.1056/NEJMcp2027084	Mycobacterium tuberculosis meningitis	Mycobacterium tuberculosis meningitis		
10.1056/NEJMcp2027087	Hereditary angioedema with C1 inhibitor deficiency and chronic spontaneous urticaria.	Hereditary angioedema with C1 inhibitor deficiency and chronic spontaneous urticaria.		Hereditary Angioedema (HAE)
10.1056/NEJMcp2027088	Diffuse idiopathic pulmonary neuroendocrine cell hyperplasia.	Diffuse idiopathic pulmonary neuroendocrine cell hyperplasia.		
10.1056/NEJMcp2027089	Lymphoplasmacytic lymphoma of the central nervous system (Bing–Neel syndrome).	Lymphoplasmacytic lymphoma of the central nervous system (Bing–Neel syndrome)	Optic nerve infiltration by Waldenström's macroglobulinemia	
10.1056/NEJMcp2027090	Normal pressure hydrocephalus.	Normal pressure hydrocephalus.		
10.1056/NEJMcp2027091	Plasma cell myeloma (multiple myeloma)	Plasma cell myeloma (multiple myeloma)		

10.1056/NEJMcpc2027093	Meningococcal purpura fulminans.	Meningococcal purpura fulminans/Purpura fulminans		
10.1056/NEJMcpc2027094	Pheochromocytoma.	Pheochromocytoma.		
10.1056/NEJMcpc2027096	Severe acquired aplastic anemia after infection with severe acute respiratory syndrome coronavirus 2.	Severe acquired aplastic anemia after infection with severe acute respiratory syndrome coronavirus 2.	Aplastic anemia	
10.1056/NEJMcpc2100271	Granulomatosis with polyangiitis.	Granulomatosis with polyangiitis.		ANCA-associated vasculitis
10.1056/NEJMcpc2100272	Delayed posthypoxic leukoencephalopathy	Delayed posthypoxic leukoencephalopathy		Hypoxic-ischemic encephalopathy/Anoxic Brain Injury
10.1056/NEJMcpc2100273	Bartonella henselae infection (cat scratch disease)	Bartonella henselae infection (cat scratch disease)		
10.1056/NEJMcpc2100274	Lactic acidosis associated with metformin use.	Lactic acidosis associated with metformin use.		Lactic acidosis and medication-induced nephrotoxicity
10.1056/NEJMcpc2100275	Babesiosis.	Babesiosis.		Tick-borne disease
10.1056/NEJMcpc2100276	Varicella–zoster virus infection.	Varicella–zoster virus infection.		
10.1056/NEJMcpc2100278	Staphylococcus aureus bacteremia and infection of a vascular graft.	Staphylococcus aureus bacteremia and infection of a vascular graft.		Infectious Aortic Aneurysm
10.1056/NEJMcpc2100281	Acquired factor VIII inhibitor.	Acquired factor VIII inhibitor/Acquired Hemophilia A		
10.1056/NEJMcpc2100282	Hereditary hemochromatosis.	Hereditary hemochromatosis.		
10.1056/NEJMcpc2103460	Cerebral amyloid angiopathy–related inflammation.	Cerebral amyloid angiopathy–related inflammation.		
10.1056/NEJMcpc2103461	Cytomegalovirus-induced	Cytomegalovirus-induced		Transplant-associated

	immune thrombocytopenia.	immune thrombocytopenia.		thrombotic microangiopathy (TA-TMA)
10.1056/NEJMcp2107344	Listeria monocytogenes bacteremia resulting in loss of fetus.	Listeria monocytogenes bacteremia resulting in loss of fetus/Listeria		
10.1056/NEJMcp2107345	Acute human immunodeficiency virus type 1 infection.	Acute human immunodeficiency virus type 1 infection.		
10.1056/NEJMcp2107346	Systemic juvenile idiopathic arthritis.	Systemic juvenile idiopathic arthritis.		
10.1056/NEJMcp2107347	Vascular Ehlers–Danlos syndrome.	Vascular Ehlers–Danlos syndrome.		
10.1056/NEJMcp2107348	Phosphaturic mesenchymal tumor causing osteomalacia.	Phosphaturic mesenchymal tumor causing osteomalacia.	Tumor-induced osteomalacia (TIO)	
10.1056/NEJMcp2107349	Neurosyphilis	Neurosyphilis	Syphilis	
10.1056/NEJMcp2107351	Primary hyperparathyroidism and brown tumor.	Primary hyperparathyroidism and brown tumor.		
10.1056/NEJMcp2107352	Eosinophilic leukemia.	Eosinophilic leukemia.		Paraneoplastic pemphigus associated with underlying lymphoma/Lymphoma with paraneoplastic manifestations
10.1056/NEJMcp2107353	Anti–melanoma differentiation–associated protein 5 (anti–MDA-5) dermatomyositis.	Anti–melanoma differentiation–associated protein 5 (anti–MDA-5) dermatomyositis/Dermatomyositis		
10.1056/NEJMcp2107354	Lead poisoning.	Lead poisoning.		
10.1056/NEJMcp2107355	Fat embolism syndrome.	Fat embolism syndrome.		
10.1056/NEJMcp2107356	Pneumocystis jirovecii pneumonia.	Pneumocystis jirovecii pneumonia.		
10.1056/NEJMcp2107357	Erysipelothrix rhusiopathiae	Erysipelothrix rhusiopathiae		

	infection.	infection/Erysipeloid		
10.1056/NEJMcp2107358	Moyamoya disease.	Moyamoya disease.		
10.1056/NEJMcp2115844	Anti-IgLON5 IgG– associated neurologic disorder	Anti-IgLON5 IgG– associated neurologic disorder		
10.1056/NEJMcp2115845	Amplatzer septal occluder device erosion	Amplatzer septal occluder device erosion	Iatrogenic Hemorrhagic Pericardial Effusion related to Amplatzer Septal Occluder Device/Complication from atrial septal occluder device	
10.1056/NEJMcp2115846	Anaplasmosis	Anaplasmosis		
10.1056/NEJMcp2115847	VEXAS (vacuoles, E1 enzyme, X-linked, autoinflammatory, somatic) syndrome.	VEXAS (vacuoles, E1 enzyme, X-linked, autoinflammatory, somatic) syndrome.		
10.1056/NEJMcp2115848	Immune checkpoint inhibitor–induced diabetes	Immune checkpoint inhibitor–induced diabetes		
10.1056/NEJMcp2115849	Idiopathic multicentric Castleman’s disease	Idiopathic multicentric Castleman’s disease		
10.1056/NEJMcp2115850	Inflammatory bowel disease (Crohn’s disease)	Crohn’s disease		Inflammatory bowel disease
10.1056/NEJMcp2115851	Granulomatosis with polyangiitis	Granulomatosis with polyangiitis		ANCA-associated vasculitis
10.1056/NEJMcp2115852	Genetic Creutzfeldt–Jakob disease.	Genetic Creutzfeldt–Jakob disease.	Creutzfeldt–Jakob disease	
10.1056/NEJMcp2115853	Diffuse large B-cell lymphoma.	Diffuse large B-cell lymphoma/Lymphoma.		
10.1056/NEJMcp2115854	Plasma-cell myeloma post- transplantation lymphoproliferative disorder.	Plasma-cell myeloma post- transplantation lymphoproliferative	PTLD with plasmacytic differentiation/PTLD	

		disorder(PTLD).		
10.1056/NEJMcp2115855	Systemic lupus erythematosus with antiphospholipid syndrome	Systemic lupus erythematosus with antiphospholipid syndrome		Systemic lupus erythematosus/Antiphospholipid syndrome
10.1056/NEJMcp2115856	High-grade B-cell lymphoma, not otherwise specified.	High-grade B-cell lymphoma, not otherwise specified.	Lymphoma	
10.1056/NEJMcp2115857	Cotard's syndrome, catatonia, and depression after coronavirus disease 2019.	Cotard's syndrome, catatonia, and depression after coronavirus disease 2019.		Cotard's syndrome and depression
10.1056/NEJMcp2115858	Well-differentiated pancreatic insulin-secreting neuroendocrine tumor (insulinoma)	Well-differentiated pancreatic insulin-secreting neuroendocrine tumor (insulinoma)		
10.1056/NEJMcp2201230	Fabry's disease.	Fabry's disease.		
10.1056/NEJMcp2201232	Myelodysplastic syndrome.	Myelodysplastic syndrome.		
10.1056/NEJMcp2201233	Regional myocarditis due to infection with Listeria monocytogenes	Regional myocarditis due to infection with Listeria monocytogenes	Listeria	
10.1056/NEJMcp2201234	Aspiration pneumonia	Aspiration pneumonia		
10.1056/NEJMcp2201235	Sarcoidosis.	Sarcoidosis.		
10.1056/NEJMcp2201236	Immune dysregulation, polyendocrinopathy, enteropathy, X-linked (IPEX) syndrome.	Immune dysregulation, polyendocrinopathy, enteropathy, X-linked (IPEX) syndrome.		
10.1056/NEJMcp2201237	Intentional guanfacine overdose.	Intentional guanfacine overdose.		Combined Effect of Olanzapine with Guanfacine/Clonidine
10.1056/NEJMcp2201238	Infection with Blastomyces dermatitidis.	Infection with Blastomyces dermatitidis.		

10.1056/NEJMcpc2201239	Linitis plastica (invasive gastric adenocarcinoma)	Linitis plastica (invasive gastric adenocarcinoma)		Gastric adenocarcinoma
10.1056/NEJMcpc2201240	Chronic salicylate toxicity	Chronic salicylate toxicity		
10.1056/NEJMcpc2201241	Chronic Candida albicans meningitis	Chronic Candida albicans meningitis		
10.1056/NEJMcpc2201243	Neonatal hypoglycemia due to biologically active teratoma and the Sotos syndrome.	Neonatal hypoglycemia due to biologically active teratoma and the Sotos syndrome.		Teratoma
10.1056/NEJMcpc2201244	Infection with monkeypox virus, West African clade.	Infection with monkeypox virus, West African clade/Monkeypox		
10.1056/NEJMcpc2201245	Eosinophilic myocarditis and drug reaction with eosinophilia and systemic symptoms (DRESS).	Eosinophilic myocarditis and drug reaction with eosinophilia and systemic symptoms (DRESS).		
10.1056/NEJMcpc2201246	Encephalitis due to Behçet's disease.	Encephalitis due to Behçet's disease/Behçet's disease		
10.1056/NEJMcpc2201247	Langerhans-cell histiocytosis of the lung and liver	Langerhans-cell histiocytosis of the lung and liver/Langerhans-cell histiocytosis		
10.1056/NEJMcpc2201248	Mansonella perstans infection.	Mansonella perstans infection.		Filariasis
10.1056/NEJMcpc2201249	Primary biliary cholangitis with portopulmonary hypertension.	Primary biliary cholangitis with portopulmonary hypertension.		Primary biliary cholangitis
10.1056/NEJMcpc2201250	Adrenocortical carcinoma.	Adrenocortical carcinoma.		Subclinical Cushing's syndrome/Adrenal adenoma
10.1056/NEJMcpc2211355	Meningioma, meningothelial type, World Health Organization grade 1.	Meningioma, meningothelial type, World Health Organization grade 1/Optic		

		nerve sheath meningioma		
10.1056/NEJMcp2211356	Systemic lupus erythematosus with membranous lupus nephritis. Testing of a random urine sample	Systemic lupus erythematosus with membranous lupus nephritis. Testing of a random urine sample	Systemic lupus erythematosus	
10.1056/NEJMcp2211357	Disseminated Mycobacterium tuberculosis infection.	Disseminated Mycobacterium tuberculosis infection.		
10.1056/NEJMcp2211358	Multiple vitamin deficiencies, including vitamin C deficiency	Multiple vitamin deficiencies, including vitamin C deficiency	Scurvy (Vitamin C Deficiency)	
10.1056/NEJMcp2211360	Cardiac paraganglioma with a germline mutation in the gene encoding succinate dehydrogenase subunit C (SDHC).	Cardiac paraganglioma with a germline mutation in the gene encoding succinate dehydrogenase subunit C (SDHC)/Cardiac paraganglioma		
10.1056/NEJMcp2211363	Hepatic adenoma, inflammatory type, with associated ischemic necrosis and extensive hemorrhage	Hepatic adenoma, inflammatory type, with associated ischemic necrosis and extensive hemorrhage/Hepatic adenoma with hemorrhage		
10.1056/NEJMcp2211364	Human granulocytic anaplasmosis.	Human granulocytic anaplasmosis.		
10.1056/NEJMcp2211365	Functional seizures.	Functional seizures/Psychogenic Non-Epileptic Seizures (PNES)		
10.1056/NEJMcp2211366	Cushing's syndrome due to ectopic secretion of adrenocorticotrophic hormone	Cushing's syndrome due to ectopic secretion of adrenocorticotrophic hormone		

	from small-cell lung cancer.	from small-cell lung cancer.		
10.1056/NEJMcp2211367	Catastrophic antiphospholipid syndrome	Catastrophic antiphospholipid syndrome	Antiphospholipid syndrome	
10.1056/NEJMcp2211368	Paraneoplastic vasculitis of the central nervous system.	Paraneoplastic vasculitis of the central nervous system.		Trousseau's syndrome
10.1056/NEJMcp2211369	Mucormycosis.	Mucormycosis.		
10.1056/NEJMcp2211370	Breast cancer–associated thrombotic microangiopathy.	Breast cancer–associated thrombotic microangiopathy.		
10.1056/NEJMcp2211375	Hypocalcemic myopathy due to hypoparathyroidism.	Hypocalcemic myopathy due to hypoparathyroidism.		
10.1056/NEJMcp2211421	Human immunodeficiency virus type 1 infection and syphilis.	Human immunodeficiency virus type 1 infection and syphilis.		Syphilis/HIV
10.1056/NEJMcp2211511	Mpox conjunctivitis.	Mpox conjunctivitis.	Mpox	
10.1056/NEJMcp2211512	Facioscapulohumeral muscular dystrophy	Facioscapulohumeral muscular dystrophy		
10.1056/NEJMcp2300894	Takayasu’s arteritis	Takayasu’s arteritis		
10.1056/NEJMcp2300895	Functional vitamin B12 deficiency from use of nitrous oxide.	Functional vitamin B12 deficiency from use of nitrous oxide.	Nitrous oxide-induced neuropathy	
10.1056/NEJMcp2300896	Factitious disorder.	Factitious disorder.		
10.1056/NEJMcp2300898	Kounis syndrome due to exposure to piperacillin–tazobactam	Kounis syndrome due to exposure to piperacillin–tazobactam		Anaphylaxis to piperacillin-tazobactam and coronary vasospasm
10.1056/NEJMcp2300899	Doege–Potter syndrome	Doege–Potter syndrome	Non-islet cell tumor hypoglycemia (NICTH) associated with metastatic solitary fibrous tumor	
10.1056/NEJMcp2300903	Turner syndrome.	Turner syndrome.		
10.1056/NEJMcp2300904	Systemic immunoglobulin light-chain (AL)	Systemic immunoglobulin light-chain (AL)	Amyloidosis	

	amyloidosis.	amyloidosis.		
10.1056/NEJMcp2300906	Congenital absence of the main portal vein with a congenital extrahepatic portosystemic shunt (Abernethy malformation).	Congenital absence of the main portal vein with a congenital extrahepatic portosystemic shunt (Abernethy malformation).		Congenital portosystemic shunt with secondary pulmonary hypertension
10.1056/NEJMcp2300907	Noncirrhotic hyperammonemia after Roux-en-Y gastric bypass	Noncirrhotic hyperammonemia after Roux-en-Y gastric bypass		
10.1056/NEJMcp2300908	Carcinoid heart disease due to a metastatic welldifferentiated neuroendocrine tumor (carcinoid tumor) associated with carcinoid syndrome.	Carcinoid heart disease due to a metastatic welldifferentiated neuroendocrine tumor (carcinoid tumor) associated with carcinoid syndrome	Carcinoid heart disease	
10.1056/NEJMcp2300968	Granulomatous lung disease as an extraintestinal manifestation of inflammatory bowel disease.	Granulomatous lung disease as an extraintestinal manifestation of inflammatory bowel disease.		
10.1056/NEJMcp2300973	Limb-shaking transient ischemic attacks.	Limb-shaking transient ischemic attacks.	Transient ischemic attacks.	
10.1056/NEJMcp2300974	BRAF–MEK inhibitor–related toxic effects.	BRAF–MEK inhibitor–related toxic effects.		
10.1056/NEJMcp2301032	Cronkhite–Canada syndrome.	Cronkhite–Canada syndrome.		
10.1056/NEJMcp2309349	Chorea due to antiphospholipid syndrome and underlying systemic lupus erythematosus.	Chorea due to antiphospholipid syndrome and underlying systemic lupus erythematosus/Antiphospholipid syndrome and systemic lupus erythematosus.		Antiphospholipid syndrome

10.1056/NEJMcpc2309382	Plasmodium falciparum malaria.	Plasmodium falciparum malaria.		malaria
10.1056/NEJMcpc2309497	Campylobacter fetus bacteremia and osteomyelitis.	Campylobacter fetus bacteremia and osteomyelitis.		Osteomyelitis
10.1056/NEJMcpc2309498	Vitamin B12 deficiency due to pernicious anemia	Vitamin B12 deficiency due to pernicious anemia		
10.1056/NEJMcpc2309725	Infant botulism	Infant botulism		
10.1056/NEJMcpc2312724	Subacute thyroiditis	Subacute thyroiditis		
10.1056/NEJMcpc2312725	Rheumatoid arthritis with vasculitis causing a confluent mononeuritis multiplex.	Rheumatoid arthritis with vasculitis causing a confluent mononeuritis multiplex.		Rheumatoid arthritis
10.1056/NEJMcpc2312728	Thyrotoxic periodic paralysis associated with Graves' disease.	Thyrotoxic periodic paralysis associated with Graves' disease.		
10.1056/NEJMcpc2312729	Type B insulin resistance syndrome.	Type B insulin resistance syndrome/Insulin Receptor Antibody-Mediated Autoimmune Insulin Resistance		Type A Insulin Resistance Syndrome
10.1056/NEJMcpc2312731	Staphylococcus aureus bacteremia and infectious aortic pseudoaneurysm.	Staphylococcus aureus bacteremia and infectious aortic pseudoaneurysm.		
10.1056/NEJMcpc2312732	MIDD (maternally inherited diabetes and deafness) and MELAS (mitochondrial encephalopathy, lactic acidosis, and strokelike episodes) associated with the m.3243A→G pathogenic variant.	MIDD (maternally inherited diabetes and deafness) and MELAS (mitochondrial encephalopathy, lactic acidosis, and strokelike episodes) associated with the m.3243A→G pathogenic variant.		MELAS

10.1056/NEJMcp2312740	Myeloperoxidase antineutrophil cytoplasmic antibody–associated vasculitis.	Myeloperoxidase antineutrophil cytoplasmic antibody–associated vasculitis.		Granulomatosis with polyangiitis
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Figure S1: Heatmaps of diagnostic performance comparison among Different LLMs using various prompts in single-round sampling

a, b. The heatmaps showing significant differences in diagnostic accuracy and coverage respectively among five LLMs using various prompts in single-round sampling for the NEJM dataset.

c, d. The heatmaps showing significant differences in diagnostic accuracy and coverage respectively among five LLMs using various prompts in single-round sampling for the offline dataset.

a)

Heatmap showing the coverage of 15 different models across 15 different scenarios. The models are listed on the left and top, and the scenarios are listed on the right. The color scale represents the coverage percentage, ranging from 0 (blue) to 1 (red).

Models (Left):

- Antony (V)
- Antony Only Class-1.5 Summ
- Antony Only Class-3 Ops
- Antony Only CPT-4s
- Antony Only CPT-4.5
- Antony Only CPT-4.5 + 1.5 Pro
- AKR Class-1.5 Summ
- AKR CPT-4s
- AKR CPT-4.5
- AKR CPT-4.5 + 1.5 Pro
- Cvt Class-3 Summ
- Cvt CPT-4s
- Cvt CPT-4.5
- Cvt CPT-4.5 + 1.5 Pro

Scenarios (Right):

- Antony (V)
- Antony Only Class-1.5 Summ
- Antony Only Class-3 Ops
- Antony Only CPT-4s
- Antony Only CPT-4.5
- Antony Only CPT-4.5 + 1.5 Pro
- AKR Class-1.5 Summ
- AKR CPT-4s
- AKR CPT-4.5
- AKR CPT-4.5 + 1.5 Pro
- Cvt Class-3 Summ
- Cvt CPT-4s
- Cvt CPT-4.5
- Cvt CPT-4.5 + 1.5 Pro

Color Scale (Right):

- 0 (X axis Superior)
- 0.01
- 0.05
- 0.1
- 0.2
- 1
- 0.2
- 0.1
- 0.05
- 0.01
- 0 (Y axis Superior)

[illegible]

c)

	Average (%)	ABR Only Class-1.5 Sensor	ABR Only Class-1 Ops	ABR Only CPT-4s	ABR Only CPT-3.5	ABR Only Class-1.5 Pro	ABR Class-1.5 Sensor	ABR Class-1.5 Ops	ABR CPT-4s	ABR CPT-3.5	ABR Class-1.5 Pro	CxT Class-1.5 Sensor	CxT Class-1.5 Ops	CxT CPT-4s	CxT CPT-3.5	CxT Class-1.5 Pro
(Dimension)	51.2	47.3	23.4	23.4	11.4	27.9	53.2	49.3	26.4	10.9	25.4	53.2	42.3	29.4	15.4	27.9
ABR Only Class-1.5 Sensor	51.2	1.000	0.074	<0.001	<0.001	<0.001	0.414	0.527	<0.001	<0.001	<0.001	0.433	0.007	<0.001	<0.001	<0.001
ABR Only Class-1 Ops	47.3	0.074	1.000	<0.001	<0.001	<0.001	0.040	0.537	<0.001	<0.001	<0.001	0.040	0.105	<0.001	<0.001	<0.001
ABR Only CPT-4s	23.4	<0.001	<0.001	1.000	<0.001	0.106	<0.001	<0.001	0.239	<0.001	0.516	<0.001	<0.001	0.011	0.008	0.136
ABR Only CPT-3.5	11.4	<0.001	<0.001	<0.001	1.000	<0.001	<0.001	<0.001	<0.001	0.763	<0.001	<0.001	<0.001	<0.001	0.088	<0.001
ABR Only Class-1.5 Pro	27.9	<0.001	<0.001	0.106	<0.001	1.000	<0.001	<0.001	0.577	<0.001	0.338	<0.001	<0.001	0.532	<0.001	1.000
ABR Class-1.5 Sensor	53.2	0.414	0.040	<0.001	<0.001	<0.001	1.000	0.228	<0.001	<0.001	1.000	0.001	<0.001	<0.001	<0.001	<0.001
ABR Class-1 Ops	49.3	0.527	0.537	<0.001	<0.001	<0.001	0.228	1.000	<0.001	<0.001	<0.001	0.238	0.027	<0.001	<0.001	<0.001
ABR CPT-4s	26.4	<0.001	<0.001	0.239	<0.001	0.577	<0.001	<0.001	1.000	<0.001	0.695	<0.001	<0.001	0.180	<0.001	0.647
ABR CPT-3.5	10.9	<0.001	<0.001	<0.001	0.763	<0.001	<0.001	<0.001	<0.001	1.000	<0.001	<0.001	<0.001	<0.001	0.061	<0.001
ABR Class-1.5 Pro	25.4	<0.001	<0.001	0.516	<0.001	0.336	<0.001	<0.001	0.695	<0.001	1.000	<0.001	<0.001	0.170	<0.001	0.466
CxT Class-1.5 Sensor	53.2	0.433	0.040	<0.001	<0.001	<0.001	1.000	0.228	<0.001	<0.001	<0.001	1.000	0.001	<0.001	<0.001	<0.001
CxT Class-1 Ops	42.3	0.007	0.105	<0.001	<0.001	<0.001	<0.001	0.023	<0.001	<0.001	0.001	1.000	<0.001	<0.001	<0.001	<0.001
CxT CPT-4s	29.4	<0.001	<0.001	0.011	<0.001	0.532	<0.001	<0.001	0.180	<0.001	0.170	<0.001	<0.001	1.000	<0.001	0.602
CxT CPT-3.5	15.4	<0.001	<0.001	0.008	0.088	<0.001	<0.001	<0.001	0.061	<0.001	<0.001	<0.001	<0.001	<0.001	1.000	<0.001
CxT Class-1.5 Pro	27.9	<0.001	<0.001	0.136	<0.001	1.000	<0.001	<0.001	0.647	<0.001	0.466	<0.001	<0.001	0.602	<0.001	1.000

P Value:

- 0 (X axis Superior)
- 0.01
- 0.05
- 0.1
- 0.2
- 1
- 0.2
- 0.1
- 0.05
- 0.01
- 0 (Y axis Superior)

d)

Coverage (%)	Average Only Class-1.5 Summ	Average Only Class-3 Ops	Average Only GPT-4s	Average Only GPT-3.5	Average Only Gemini-1.5 Pro	AAR Class-1.5 Summ	AAR Class-3 Ops	AAR GPT-4s	AAR GPT-3.5	AAR Gemini-1.5 Pro	GvT Class-1.5 Summ	GvT Class-3 Ops	GvT GPT-4s	GvT GPT-3.5	GvT Gemini-1.5 Pro
Coverage (%)	75.1	68.2	43.8	29.4	51.2	79.6	73.6	48.8	30.3	52.7	81.1	67.2	53.2	34.8	48.3
Average Only Class-1.5 Summ	75.1	1.000	-0.013	<0.001	<0.001	-0.001	0.117	0.602	<0.001	<0.001	<0.001	0.014	0.364	<0.001	<0.001
Average Only Class-3 Ops	68.2	0.013	1.000	<0.001	<0.001	<0.001	0.086	<0.001	<0.001	<0.001	<0.001	0.758	<0.001	<0.001	<0.001
Average Only GPT-4s	43.8	<0.001	<0.001	1.000	<0.001	0.039	<0.001	<0.001	0.059	<0.001	0.008	<0.001	<0.001	<0.001	0.014
Average Only GPT-3.5	29.4	<0.001	<0.001	<0.001	1.000	<0.001	<0.001	<0.001	0.739	<0.001	<0.001	<0.001	<0.001	0.109	<0.001
Average Only Gemini-1.5 Pro	51.2	<0.001	<0.001	<0.001	<0.001	1.000	<0.001	<0.001	0.492	<0.001	0.668	<0.001	<0.001	0.564	<0.001
AAR Class-1.5 Summ	79.6	0.117	<0.001	<0.001	<0.001	<0.001	1.000	0.064	<0.001	<0.001	0.590	<0.001	<0.001	<0.001	<0.001
AAR Class-3 Ops	73.6	0.602	0.086	<0.001	<0.001	<0.001	0.064	1.000	<0.001	<0.001	0.809	0.037	<0.001	<0.001	<0.001
AAR GPT-4s	48.8	<0.001	<0.001	0.059	<0.001	0.492	<0.001	<0.001	1.000	<0.001	0.228	<0.001	<0.001	0.039	0.896
AAR GPT-3.5	30.3	<0.001	<0.001	<0.001	0.739	<0.001	<0.001	<0.001	<0.001	1.000	<0.001	<0.001	<0.001	0.189	<0.001
AAR Gemini-1.5 Pro	52.7	<0.001	<0.001	0.008	<0.001	0.668	<0.001	<0.001	0.228	<0.001	1.000	<0.001	<0.001	0.881	<0.001
GvT Class-1.5 Summ	81.1	0.014	<0.001	<0.001	<0.001	<0.001	0.590	<0.001	<0.001	<0.001	<0.001	1.000	<0.001	<0.001	<0.001
GvT Class-3 Ops	67.2	0.016	0.758	<0.001	<0.001	<0.001	<0.001	0.037	<0.001	<0.001	<0.001	<0.001	1.000	<0.001	<0.001
GvT GPT-4s	53.2	<0.001	<0.001	<0.001	<0.001	0.564	<0.001	<0.001	0.139	<0.001	0.881	<0.001	<0.001	1.000	<0.001
GvT GPT-3.5	34.8	<0.001	<0.001	0.014	0.109	<0.001	<0.001	<0.001	0.189	<0.001	<0.001	<0.001	<0.001	<0.001	1.000
GvT Gemini-1.5 Pro	48.3	<0.001	<0.001	0.241	<0.001	0.439	<0.001	<0.001	0.896	<0.001	0.189	<0.001	<0.001	0.157	<0.001

P Value:

0 (X axis Superior)

0.01

0.05

0.1

0.2

1

0.2

0.1

0.05

0.01

0 (Y axis Superior)