‘Input text’: {'Case Report': "The proband was a 37-year-old man who had visual and gait disturbances that had first appeared at 10 years of age. He showed horizontal gaze palsy, gaze-evoked nystagmus, dysarthria, and cerebellar ataxia. Brain and orbit MRI disclosed atrophy of the optic nerve and cerebellum, and degen erative changes in the bilateral inferior olivary nucleus. Mutational analyses of mitochondrial DNA identifie d the coexistence of heteroplasmic G11778A and homoplasmic T3394C mutations. The proband (III-1) (Fi g. 1) was a 37-year-old man with severe dizziness and double vision. He had first experienced visual and g ait disturbances at 10 years of age. The neurological examination performed on admission revealed mild di sturbance of cognitive function (Revised Wechsler Adult Intelligence Scale: total IQ=73, performance IQ=5 8, verbal IQ=91). Neurological disturbances were observed including bilateral exotropia, double vision, inc omplete horizontal movement of the eyes to the bilateral side, horizontal, and vertical gaze-evoked nystag mus, and dysarthria. The light reflex was prompt. No disturbances in cranial nerves I, VII, VIII, and XII were detected. Tremor appeared in his neck, but other involuntary movements including palatal myoclonus wer e not observed. While his upper and lower limbs showed no paralysis, they exhibited severe cerebellar atax ia and hypotonia. No abnormal findings were detected in his deep tendon reflex and sensory system. Ophth almological examination revealed atrophy of the optic nerve, but there were no pigmentation changes of th e retina. Blood and cerebrospinal fluid analyses were normal. Ergometer exercise did not up-regulate his s erum lactate and pyruvate. Orbital MRI revealed atrophy of the optic nerve (Fig. 2A), and brain MRI disclo sed severe atrophy of the cerebellum and mild atrophy of the brain stem (Fig. 2B). The bilateral inferior ol ivary nucleus exhibited low signal intensities on T1-weighted imaging, and high signal intensities on T2-we ighted imaging, suggesting degeneration (Fig. 2C and D). The patient was diagnosed as having LHON plus olivocerebellar degeneration. Although the thyrotropin-releasing drug taltirelin did not relieve his sympto ms, adenosine triphosphate disodium reduced his dizziness. The patient's mother (II-2) and uncle (II-3) als o had optic neuropathy, but other neurological abnormalities such as ataxia and dystonia were not observe d. The patient's mother has a history of subarachnoid hemorrhage. MRI of his mother disclosed mild atrop hy of the optic nerve (Fig. 2E), pons, and cerebellum (Fig. 2F-H). No signal changes were observed in the i nferior olivary nucleus (Fig. 2F-H). We were unable to confirm the detailed clinical information of the pro band's grandmother (I-2)."

'Mutation analyses of mtDNA': "Blood samples were obtained from the patient and his mother with their i nformed consent, and the methods used were approved by the institutional review board of Tottori Univer sity Hospital. Both mtDNA and genomic DNA were extracted by standard procedures. The polymerase chai n reaction (PCR) was carried out using the primers 5'-CCTCCCTACTATGCCTAGAAGGA-3' and 5'-TTTGGGT TGTGGCTCAGTGT-3' for ND4, including 11778G analysis, and 5'-AGTTCAGACCGGAGTAATCCAG-3' and 5'-A GGGTTGTAGTAGCCCGTAG-3' for ND1 . The primer set for ND4 was designed to identify G11778A mutation s, which is the main mutation for LHON. The primer set for ND1 was designed to detect not only the T3394 C mutation as a minor mutation for LHON but also an A3243G mutation that is frequently detected in patie nts with mitochondrial encephalomyopathy, lactic acidosis, and stroke-like episodes. PCR products that inc luded the previously reported candidate abnormal points were analyzed by capillary electrophoresis using an automated DNA sequencer. The G11778A and T3394C mutations were identified, while the A3243G mu tation was not detected. The mutations in the mtDNA were confirmed by performing PCR-restriction fragm ent length polymorphism (RFLP), in which the PCR products were digested using either HaeIII (for T3394 C) or Tsp45I (for G11778A). In order to quantify the heteroplasmic mutation of G117 78A, we prepared ve ctor constructs including 11778G or 11 778A, and semiquantitative analyses of G11778A were performed using a mixture of each with several rate standards (described in Fig. 4). Sequence analysis revealed the h omoplasmic T3394C mutation of the mtDNA (Fig. 3A). This mutation causes a Tyr-to-His amino acid subst itution in ND1. PCR-RFLP data revealed that the T3394C mutation present in both the proband (III-1) and his mother (II-2) was homoplasmic; differences between the patient and his mother were not observed for this mutation (Fig. 3B). The G11778A mutation, which causes an Arg-to-His amino acid substitution in ND 4, was also observed (Fig. 4A). PCR-RFLP data showed that this mutation in the patient and his mother wa s heteroplasmic (Fig. 4B). The semiquantitative analysis performed to determine the effect of this mutatio n on disease severity revealed that III-1 had a 92% heteroplasmic G11778A mutation, and II-2 had a 70% h eteroplasmic G11778A mutation (Fig. 4C). Established genetic abnormalities associated with cerebellar at axia including polyglutamine diseases were not found."}

Model and Prompting Strategies	Example LLM Extracted Preprocessed Output
Llama3:8B-Instruct Few-Shot Prompts Category-Based Filtering and Targeted Extraction	{'iem': ['iem: Inborn errors of metabolism'], 'Vitals_Hema': [], 'Pregnancy': ['maternal_health: Subarachnoid hemorrhage'], 'Neuro': ['neurological: Horizontal gaze palsy', 'neurological: Gaze-evoked nystagmus', 'neurological: Dysarthria', 'neurological: Cerebellar ataxia', 'neurological: Bilateral exotropia', 'neurological: Double vision', 'neurological: Incomplete horizontal movement of the eyes to the bilateral side', 'neurological: Horizontal and vertical gaze-evoked nystagmus', 'neurological: Dysarthria', 'neurological: Severe cerebellar ataxia', 'neurological: Hypotonia', 'cognitive: Mild disturbance of cognitive function (Revised Wechsler Adult Intelligence Scale: total IQ=73, performance IQ=58, verbal IQ=91)'], 'CVS': [], 'RESP': [], 'EENT': ['eyes: bilateral exotropia', 'eyes: double vision', 'eyes: incomplete horizontal movement of the eyes to the bilateral side', 'eyes: horizontal and vertical gaze-evoked nystagmus'], 'GI': [], 'GU': [], 'DERM': [], 'MSK': ['muscle: Severe cerebellar ataxia and hypotonia', 'muscle: Tremor appeared in his neck', 'skeletal: Atrophy of the optic nerve', 'skeletal: Mild atrophy of the brain stem', 'skeletal: Severe atrophy of the cerebellum'], 'ENDO': [], 'LYMPH': [], 'History': ['history_of_present_illness: Visual and gait disturbances first appeared at 10 years of age, with gradual onset of symptoms', "family_and_genetics_history: Proband's mother (II-2) had optic neuropathy", "family_and_genetics_history: Proband's uncle (II-3) also had optic neuropathy", "family_and_genetics_history: Proband's grandmother (I-2)'s clinical information was unavailable", 'chief_complaint: Severe dizziness and double vision'], 'Lab_Image': ['Lab_Image: Brain and orbit MRI disclosed atrophy of the optic nerve and cerebellum, and degenerative changes in the bilateral inferior olivary nucleus', 'Lab_Image: Orbital MRI revealed atrophy of the optic nerve (Fig. 2A)', 'Lab_Image: Brain MRI disclosed severe atrophy of the cerebellum and mild atrophy of the brain stem (Fig. 2B)', 'Lab_Image: The bilateral inferior olivary nucleus exhibited low signal intensities on T1-weighted imaging, and high signal intensities on T2-weighted imaging, suggesting degeneration (Fig. 2C and D)']}

Llama3:8B-Instruct Few-shot prompts Uniform Application of Category Prompts	{'iem': ['iem: Inborn errors of metabolism'], 'Vitals_Hema': [], 'Pregnancy': ['maternal_health: Subarachnoid hemorrhage'], 'Neuro': ['neurological: Horizontal gaze palsy', 'neurological: Gaze-evoked nystagmus', 'neurological: Dysarthria', 'neurological: Cerebellar ataxia', 'neurological: Bilateral exotropia', 'neurological: Double vision', 'neurological: Incomplete horizontal movement of the eyes to the bilateral side', 'neurological: Horizontal and vertical gaze-evoked nystagmus', 'neurological: Dysarthria', 'neurological: Severe cerebellar ataxia', 'neurological: Hypotonia', 'cognitive: Mild disturbance of cognitive function (Revised Wechsler Adult Intelli gence Scale: total IQ=73, performance IQ=58, verbal IQ=91)'], 'CVS': [], 'RESP': [], 'EENT': ['eyes: bilateral exotropia', 'eyes: double vision', 'eyes: incomplete horizontal movement of the eyes to the bilateral side', 'eyes: horizontal and vertical gaze-evoked nystagmus'], 'GI': [], 'GU': [], 'DERM': [], 'MSK': ['muscle: Severe cerebellar ataxia and hypotonia', 'muscle: Tremor appeared in his neck', 'skeletal: Atrophy of the optic nerve', 'skeletal: Mild atrophy of the brain stem', 'skeletal: Severe atrophy of the cerebellum'], 'ENDO': [], 'LYMPH': ['bone_marrow: Bone marrow biopsy shows increased cellularity'], 'History': ['history_of_present_illness: Visual and gait disturbances first appeared at 10 years of age, with gradual onset of symptoms', "family_and_genetics_history: Proband's mother (II-2) had optic neuropathy", "family_and_genetics_history: Proband's uncle (II-3) also had optic neuropathy", "family_and_genetics_history: Proband's grandmother (I-2)'s clinical informatio n was unavailable", 'chief_complaint: Severe dizziness and double vision', 'family_and_genetics_history: Father had colon cancer', 'family_and_genetics_history: Sister diagnosed with breast cancer at age 50'], 'Lab_Image': ['Lab_Image: Brain and orbit MRI disclosed atrophy of the optic nerv e and cerebellum, and degenerative changes in the bilateral inferior olivary nucle us', 'Lab_Image: Orbital MRI revealed atrophy of the optic nerve (Fig. 2A)', 'Lab_Image: Brain MRI disclosed severe atrophy of the cerebellum and mild atro phy of the brain stem (Fig. 2B)', 'Lab_Image: The bilateral inferior olivary nucleus exhibited low signal intensities on T1-weighted imaging, and high signal intensities on T2-weighted imaging, sug gesting degeneration (Fig. 2C and D)', 'Lab_Image: Sequence analysis revealed the homoplasmic T3394C mutation of th e mtDNA (Fig. 3A). This mutation causes a Tyr-to-His amino acid substitution in N D1.',

	'Lab_Image: PCR-RFLP data revealed that the T3394C mutation present in both t he proband (III-1) and his mother (II-2) was homoplasmic; differences between t he patient and his mother were not observed for this mutation (Fig. 3B).', 'Lab_Image: The G11778A mutation, which causes an Arg-to-His amino acid subs titution in ND4, was also observed (Fig. 4A). PCR-RFLP data showed that this mut ation in the patient and his mother was heteroplasmic (Fig. 4B). The semiquantita tive analysis performed to determine the effect of this mutation on disease severit y revealed that III-1 had a 92% heteroplasmic G11778A mutation, and II-2 had a 70% heteroplasmic G11778A mutation (Fig. 4C).', 'Lab_Image: Sequence analysis revealed the homoplasmic T3394C mutation of th e mtDNA', 'Lab_Image: PCR-RFLP data revealed that the T3394C mutation present in both t he proband (III-1) and his mother (II-2) was homoplasmic', 'Lab_Image: The G11778A mutation, which causes an Arg-to-His amino acid subs titution in ND4, was also observed', 'Lab_Image: PCR-RFLP data showed that this mutation in the patient and his mot her was heteroplasmic']}
Llama3:8B-Instruct Few-Shot Prompt System-Wide Unified Extraction	{'IEM': ['IEM: is_IEM'], 'pregnancy': ['pregnancy: neonatal_health', 'pregnancy: maternal_health', 'pregnancy: pregnancy_lab_tests_imaging_exam'], 'Vitals_Hema': ['Vitals_Hema: temperature', 'Vitals_Hema: pulse', 'Vitals_Hema: respiratory_rate', 'Vitals_Hema: blood_pressure', 'Vitals_Hema: oxygen_saturation', 'Vitals_Hema: hematological_conditions', 'Vitals_Hema: hematology_lab_tests_measurements'], 'Neuro': ['Neuro: neurological', 'Neuro: cognitive', 'Neuro: neuro_lab_tests_imaging_exam'], 'EENT': ['EENT: eyes', 'EENT: ears', 'EENT: nose', 'EENT: throat', 'EENT: EENT_lab_tests_imaging_exam'], 'CVS': ['CVS: cardiac', 'CVS: vascular', 'CVS: CVS_lab_tests_imaging_exam'], 'RESP': ['RESP: respiratory', 'RESP: respiratory_system_lab_tests_imaging_exam'], 'GI': ['GI: genital', 'GI: urinary', 'GI: GU_lab_tests_imaging_exam'], 'ENDO': ['ENDO: endocrine_glands', 'ENDO: Endocrine_lab_tests_image_exam'], 'History': ['History: past_medical_history', 'History: past_surgical_history', 'History: history_of_present_illness', 'History: social_history', 'History: family_and_genetics_history', 'History: chief_complaint'], 'Lab_Image': []}

Llama3:8B-Instruct Zero-Shot Prompts Category-Based Filtering and Targeted Extraction	{'iem': ['iem: is_IEM'], 'Vitals_Hema': ['hematological_conditions: Optic neuropathy', 'hematological_conditions: Atrophy of the optic nerve', 'hematological_conditions: Cerebellar ataxia', 'hematological_conditions: Hypotonia'], 'Pregnancy': ['subarachnoid hemorrhage: True'], 'Neuro': ['neurological: horizontal gaze palsy', 'neurological: gaze-evoked nystagmus', 'neurological: dysarthria', 'neurological: cerebellar ataxia', 'neurological: bilateral exotropia', 'neurological: double vision', 'neurological: incomplete horizontal movement of the eyes to the bilateral side', 'neurological: horizontal and vertical gaze-evoked nystagmus', 'neurological: dysarthria', 'neurological: tremor in his neck', 'neurological: severe cerebellar ataxia and hypotonia', 'cognitive: mild disturbance of cognitive function (Revised Wechsler Adult Intelligence Scale: total IQ=73, performance IQ=58, verbal IQ=91)'], 'CVS': [], 'RESP': [], 'EENT': ['eyes: horizontal gaze palsy', 'eyes: gaze-evoked nystagmus', 'eyes: bilateral exotropia', 'eyes: double vision', 'eyes: incomplete horizontal movement of the eyes to the bilateral side', 'eyes: horizontal and vertical gaze-evoked nystagmus', 'throat: dysarthria'], 'GI': [], 'GU': [], 'DERM': ['facial_features: bilateral exotropia', 'facial_features: double vision', 'facial_features: horizontal and vertical gaze-evoked nystagmus'], 'MSK': ['muscle: Severe cerebellar ataxia and hypotonia', 'muscle: Tremor appeared in his neck', 'skeletal: Atrophy of the optic nerve', 'skeletal: Mild atrophy of the brain stem', 'skeletal: Severe atrophy of the cerebellum'], 'ENDO': [], 'LYMPH': [], 'History': ['history_of_present_illness: Visual and gait disturbances first appeared at 10 years of age', 'history_of_present_illness: Horizontal gaze palsy, gaze-evoked nystagmus, dysarthria, and cerebellar ataxia were observed', 'history_of_present_illness: Severe dizziness and double vision were reported', 'family_and_genetics_history: Mother (II-2) had optic neuropathy', 'family_and_genetics_history: Uncle (II-3) also had optic neuropathy', "family_and_genetics_history: Grandmother (I-2)'s clinical information was not confirmed", 'family_and_genetics_history: Mother has a history of subarachnoid hemorrhage', 'chief complaint: Severe dizziness and double vision'], 'Lab_Image': ['Lab_Image: Brain and orbit MRI disclosed atrophy of the optic nerve and cerebellum, and degenerative changes in the bilateral inferior olivary nucleus',

	'Lab_Image: Orbital MRI revealed atrophy of the optic nerve (Fig. 2A)', 'Lab_Image: Brain MRI disclosed severe atrophy of the cerebellum and mild atrophy of the brain stem (Fig. 2B)', 'Lab_Image: The bilateral inferior olivary nucleus exhibited low signal intensities on T1-weighted imaging, and high signal intensities on T2-weighted imaging, suggesting degeneration (Fig. 2C and D)', 'Lab_Image: MRI of his mother disclosed mild atrophy of the optic nerve (Fig. 2E), pons, and cerebellum (Fig. 2F-H)', 'Lab_Image: Orbital MRI revealed atrophy of the optic nerve (Fig. 2A)', 'Lab_Image: Brain MRI disclosed severe atrophy of the cerebellum and mild atrophy of the brain stem (Fig. 2B)', 'Lab_Image: The bilateral inferior olivary nucleus exhibited low signal intensities on T1-weighted imaging, and high signal intensities on T2-weighted imaging, suggesting degeneration (Fig. 2C and D)']}
Llama3:8B-Instruct Zero-shot prompts Uniform Application of Category Prompts	{'iem': ['iem: is_IEM'], 'Vitals_Hema': ['hematological_conditions: Optic neuropathy', 'hematological_conditions: Atrophy of the optic nerve', 'hematological_conditions: Cerebellar ataxia', 'hematological_conditions: Hypotonia'], 'Pregnancy': ['subarachnoid hemorrhage: True'], 'Neuro': ['neurological: horizontal gaze palsy', 'neurological: gaze-evoked nystagmus', 'neurological: dysarthria', 'neurological: cerebellar ataxia', 'neurological: bilateral exotropia', 'neurological: double vision', 'neurological: incomplete horizontal movement of the eyes to the bilateral side', 'neurological: horizontal and vertical gaze-evoked nystagmus', 'neurological: dysarthria', 'neurological: tremor in his neck', 'neurological: severe cerebellar ataxia and hypotonia', 'cognitive: mild disturbance of cognitive function (Revised Wechsler Adult Intelligence Scale: total IQ=73, performance IQ=58, verbal IQ=91)', 'mutation: T3394C', 'description: Causes a Tyr-to-His amino acid substitution in ND1', 'mutation: G11778A', 'description: Causes an Arg-to-His amino acid substitution in ND4, heteroplasmic in the patient and his mother'], 'CVS': ['stroke_like_episodes: True'], 'RESP': [], 'EENT': ['eyes: horizontal gaze palsy', 'eyes: gaze-evoked nystagmus', 'eyes: bilateral exotropia', 'eyes: double vision', 'eyes: incomplete horizontal movement of the eyes to the bilateral side', 'eyes: horizontal and vertical gaze-evoked nystagmus', 'throat: dysarthria'], 'GI': [], 'GU': [], 'DERM': ['facial_features: bilateral exotropia', 'facial_features: double vision', 'facial_features: horizontal and vertical gaze-evoked nystagmus'], 'MSK': ['muscle: Severe cerebellar ataxia and hypotonia', 'muscle: Tremor appeared in his neck',

	'skeletal: Atrophy of the optic nerve', 'skeletal: Mild atrophy of the brain stem', 'skeletal: Severe atrophy of the cerebellum'], 'ENDO': [], 'LYMPH': [], 'History': ['history_of_present_illness: Visual and gait disturbances first appeared at 10 years of age', 'history_of_present_illness: Horizontal gaze palsy, gaze-evoked nystagmus, dysarthria, and cerebellar ataxia were observed', 'history_of_present_illness: Severe dizziness and double vision were reported', 'family_and_genetics_history: Mother (II-2) had optic neuropathy', 'family_and_genetics_history: Uncle (II-3) also had optic neuropathy', "family_and_genetics_history: Grandmother (I-2)'s clinical information was not confirmed", 'family_and_genetics_history: Mother has a history of subarachnoid hemorrhage', 'chief complaint: Severe dizziness and double vision'], 'Lab_Image': ['Lab_Image: Brain and orbit MRI disclosed atrophy of the optic nerve and cerebellum, and degenerative changes in the bilateral inferior olivary nucleus', 'Lab_Image: Orbital MRI revealed atrophy of the optic nerve (Fig. 2A)', 'Lab_Image: Brain MRI disclosed severe atrophy of the cerebellum and mild atrophy of the brain stem (Fig. 2B)', 'Lab_Image: The bilateral inferior olivary nucleus exhibited low signal intensities on T1-weighted imaging, and high signal intensities on T2-weighted imaging, suggesting degeneration (Fig. 2C and D)', 'Lab_Image: MRI of his mother disclosed mild atrophy of the optic nerve (Fig. 2E), pons, and cerebellum (Fig. 2F-H)', 'Lab_Image: Orbital MRI revealed atrophy of the optic nerve (Fig. 2A)', 'Lab_Image: Brain MRI disclosed severe atrophy of the cerebellum and mild atrophy of the brain stem (Fig. 2B)', 'Lab_Image: The bilateral inferior olivary nucleus exhibited low signal intensities on T1-weighted imaging, and high signal intensities on T2-weighted imaging, suggesting degeneration (Fig. 2C and D)']}
Llama3:8B-Instruct Zero-shot prompts System-Wide Unified Extraction	{'Lab_Image': []}
QWEN2:7B-Instruct Few-Shot Prompts Category-Based Filtering and Targeted Extraction	{'iem': ['iem: Inborn errors of metabolism'], 'Vitals_Hema': ['hematological_conditions: Diagnosed with LHON plus olivocerebellar degeneration'], 'Pregnancy': [], 'Neuro': ['neurological: Horizontal gaze palsy', 'neurological: Gaze-evoked nystagmus', 'neurological: Dysarthria', 'neurological: Cerebellar ataxia', 'neurological: Severe cerebellar ataxia and hypotonia', 'neurological: Atrophy of the optic nerve', 'neurological: Degenerative changes in the bilateral inferior olivary nucleus', 'neurological: Bilateral exotropia',

	'neurological: Double vision', 'neurological: Incomplete horizontal movement of the eyes to the bilateral side', 'neurological: Horizontal and vertical gaze-evoked nystagmus', 'neurological: Tremor in neck', 'neurological: No disturbances in cranial nerves I, VII, VIII, and XII', 'neurological: Severe atrophy of the cerebellum', 'neurological: Mild atrophy of the brain stem', 'neurological: Low signal intensities on T1-weighted imaging for bilateral inferio r olivary nucleus', 'neurological: High signal intensities on T2-weighted imaging for bilateral inferio r olivary nucleus suggesting degeneration', 'cognitive: Mild disturbance of cognitive function (Revised Wechsler Adult Intelli gence Scale: total IQ=73, performance IQ=58, verbal IQ=91)'], 'CVS': ['vascular: Visible atrophy of the optic nerve', 'vascular: Severe cerebellar ataxia and hypotonia'], 'EENT': ['eyes: Atrophy of the optic nerve'], 'GI': [], 'GU': [], 'DERM': [], 'MSK': ['muscle: Severe cerebellar ataxia and hypotonia observed', 'skeletal: Atrophy of the optic nerve noted', 'skeletal: Severe atrophy of the cerebellum disclosed', 'skeletal: Mild atrophy of the brain stem observed', 'skeletal: Low signal intensities on T1-weighted imaging in bilateral inferior oliv ary nucleus suggesting degeneration', 'skeletal: High signal intensities on T2-weighted imaging in bilateral inferior oliv ary nucleus'], 'ENDO': [], 'LYMPH': ['lymphatic_tissues: Atrophy of the optic nerve'], 'History': ['condition: Diagnosed with LHON plus olivocerebellar degeneration', 'first_symptom: First experienced visual and gait disturbances at 10 years of age ', 'symptoms: Severe dizziness and double vision', 'symptoms: Mild disturbance of cognitive function (Revised Wechsler Adult Intel ligence Scale: total IQ=73, performance IQ=58, verbal IQ=91)', 'symptoms: Neurological disturbances including bilateral exotropia, double visio n, incomplete horizontal movement of the eyes to the bilateral side, horizontal an d vertical gaze-evoked nystagmus, dysarthria', 'symptoms: No disturbances in cranial nerves I, VII, VIII, and XII', 'symptoms: Severe cerebellar ataxia and hypotonia', 'symptoms: No abnormal findings in deep tendon reflex and sensory system', 'symptoms: Atrophy of the optic nerve observed during ophthalmological exami nation', 'symptoms: Normal blood and cerebrospinal fluid analyses', 'symptoms: No up-regulation of serum lactate and pyruvate during ergometer ex ercise', 'symptoms: MRI revealed atrophy of the optic nerve, severe atrophy of the cereb ellum and mild atrophy of the brain stem', 'symptoms: Bilateral inferior olivary nucleus exhibited low signal intensities on T1-weighted imaging and high signal intensities on T2-weighted imaging suggesti ng degeneration', 'condition: optic neuropathy', 'relation: mother', 'relation: uncle', 'condition: subarachnoid hemorrhage',

	'relation: mother', 'description: No other neurological abnormalities such as ataxia or dystonia obse rved in mother and uncle', 'condition: MRI disclosed mild atrophy of the optic nerve, pons, and cerebellum', 'relation: mother', 'description: No signal changes were observed in inferior olivary nucleus during MRI examination'], 'Lab_Image': ['Lab_Image: MRI Brain: Atrophy of the optic nerve', 'Lab_Image: MRI Brain: Severe atrophy of the cerebellum and mild atrophy of th e brain stem', 'Lab_Image: MRI Brain: Low signal intensities on T1-weighted imaging for bilate ral inferior olivary nucleus', 'Lab_Image: MRI Brain: High signal intensities on T2-weighted imaging for bilate ral inferior olivary nucleus suggesting degeneration', 'Lab_Image: MRI disclosed atrophy of the optic nerve, cerebellum, and bilateral i nferior olivary nucleus', 'Lab_Image: MRI of mother: Mild atrophy of the optic nerve', 'Lab_Image: MRI of mother: Pons and cerebellum atrophy', 'Lab_Image: MRI of mother: No signal changes in the inferior olivary nucleus', 'Lab_Image: Polymerase chain reaction (PCR) analysis: Identification of G11778 A and T3394C mutations in mtDNA', 'Lab_Image: Capillary electrophoresis using an automated DNA sequencer: Analy sis of PCR products including candidate abnormal points', 'Lab_Image: PCR-restriction fragment length polymorphism (RFLP): Confirmatio n of mutations by digesting PCR products with HaeIII or Tsp45I', 'Lab_Image: Semiquantitative analysis: Determination of the effect of G11778A mutation on disease severity', 'Lab_Image: MRI: Atrophy of the optic nerve', 'Lab_Image: MRI: Severe atrophy of the cerebellum', 'Lab_Image: MRI: Mild atrophy of the brain stem', 'Lab_Image: MRI: Low signal intensities on T1-weighted imaging in bilateral infe rior olivary nucleus (suggesting degeneration)', 'Lab_Image: MRI: High signal intensities on T2-weighted imaging in bilateral infe rior olivary nucleus (suggesting degeneration)', 'Lab_Image: MRI: Atrophy of optic nerve and cerebellum, degenerative changes i n bilateral inferior olivary nucleus', 'Lab_Image: MRI: Atrophy of the optic nerve', 'Lab_Image: MRI: Severe atrophy of the cerebellum', 'Lab_Image: MRI: Mild atrophy of the brain stem', 'Lab_Image: MRI: Low signal intensities on T1-weighted imaging in bilateral infe rior olivary nucleus (suggesting degeneration)', 'Lab_Image: MRI: High signal intensities on T2-weighted imaging in bilateral infe rior olivary nucleus (suggesting degeneration)']}

QWEN2:7B-Instruct Few-shot prompts Uniform Application of Category Prompts	{'iem': ['iem: Inborn errors of metabolism'], 'Vitals_Hema': ['hematological_conditions: Diagnosed with LHON plus olivocerebellar degeneration', 'hematological_conditions: Diagnosed with anemia'], 'Pregnancy': [], 'Neuro': ['neurological: Horizontal gaze palsy', 'neurological: Gaze-evoked nystagmus', 'neurological: Dysarthria', 'neurological: Cerebellar ataxia', 'neurological: Severe cerebellar ataxia and hypotonia', 'neurological: Atrophy of the optic nerve', 'neurological: Degenerative changes in the bilateral inferior olivary nucleus', 'neurological: Bilateral exotropia', 'neurological: Double vision', 'neurological: Incomplete horizontal movement of the eyes to the bilateral side', 'neurological: Horizontal and vertical gaze-evoked nystagmus', 'neurological: Tremor in neck', 'neurological: No disturbances in cranial nerves I, VII, VIII, and XII', 'neurological: Severe atrophy of the cerebellum', 'neurological: Mild atrophy of the brain stem', 'neurological: Low signal intensities on T1-weighted imaging for bilateral inferior olivary nucleus', 'neurological: High signal intensities on T2-weighted imaging for bilateral inferior olivary nucleus suggesting degeneration', 'cognitive: Mild disturbance of cognitive function (Revised Wechsler Adult Intelligence Scale: total IQ=73, performance IQ=58, verbal IQ=91)', 'neurological: G11778A mutation identified', 'neurological: T3394C mutation identified', 'neurological: Heteroplasmic G11778A mutation in patient and mother', 'neurological: Homoplasmic T3394C mutation in patient and mother'], 'CVS': ['vascular: Visible atrophy of the optic nerve', 'vascular: Severe cerebellar ataxia and hypotonia'], 'RESP': [], 'EENT': ['eyes: Atrophy of the optic nerve'], 'GI': [], 'GU': [], 'DERM': [], 'MSK': ['muscle: Severe cerebellar ataxia and hypotonia observed', 'skeletal: Atrophy of the optic nerve noted', 'skeletal: Severe atrophy of the cerebellum disclosed', 'skeletal: Mild atrophy of the brain stem observed', 'skeletal: Low signal intensities on T1-weighted imaging in bilateral inferior olivary nucleus suggesting degeneration', 'skeletal: High signal intensities on T2-weighted imaging in bilateral inferior olivary nucleus'], 'ENDO': [], 'LYMPH': ['lymphatic_tissues: Atrophy of the optic nerve'], 'History': ['condition: Diagnosed with LHON plus olivocerebellar degeneration', 'first_symptom: First experienced visual and gait disturbances at 10 years of age', 'symptoms: Severe dizziness and double vision', 'symptoms: Mild disturbance of cognitive function (Revised Wechsler Adult Intelligence Scale: total IQ=73, performance IQ=58, verbal IQ=91)',

	'symptoms: Neurological disturbances including bilateral exotropia, double visio n, incomplete horizontal movement of the eyes to the bilateral side, horizontal an d vertical gaze-evoked nystagmus, dysarthria', 'symptoms: No disturbances in cranial nerves I, VII, VIII, and XII', 'symptoms: Severe cerebellar ataxia and hypotonia', 'symptoms: No abnormal findings in deep tendon reflex and sensory system', 'symptoms: Atrophy of the optic nerve observed during ophthalmological exami nation', 'symptoms: Normal blood and cerebrospinal fluid analyses', 'symptoms: No up-regulation of serum lactate and pyruvate during ergometer ex ercise', 'symptoms: MRI revealed atrophy of the optic nerve, severe atrophy of the cereb ellum and mild atrophy of the brain stem', 'symptoms: Bilateral inferior olivary nucleus exhibited low signal intensities on T1-weighted imaging and high signal intensities on T2-weighted imaging suggesti ng degeneration', 'condition: optic neuropathy', 'relation: mother', 'relation: uncle', 'condition: subarachnoid hemorrhage', 'relation: mother', 'description: No other neurological abnormalities such as ataxia or dystonia obse rved in mother and uncle', 'condition: MRI disclosed mild atrophy of the optic nerve, pons, and cerebellum', 'relation: mother', 'description: No signal changes were observed in inferior olivary nucleus during MRI examination', 'history_of_present_illness: Patient was brought to ER after the first episode of ac ute chest pain and hemoptysis', 'family_and_genetics_history: Father had colon cancer', 'family_and_genetics_history: Sister diagnosed with breast cancer at age 50'], 'Lab_Image': ['Lab_Image: Hemoglobin: 13.5 g/dL', 'Lab_Image: WBC count: 6,000 /μL', 'Lab_Image: Platelet count: 250,000 /μL', 'Lab_Image: MRI Brain: Atrophy of the optic nerve', 'Lab_Image: MRI Brain: Severe atrophy of the cerebellum and mild atrophy of th e brain stem', 'Lab_Image: MRI Brain: Low signal intensities on T1-weighted imaging for bilate ral inferior olivary nucleus', 'Lab_Image: MRI Brain: High signal intensities on T2-weighted imaging for bilate ral inferior olivary nucleus suggesting degeneration', 'Lab_Image: MRI disclosed atrophy of the optic nerve, cerebellum, and bilateral i nferior olivary nucleus', 'Lab_Image: Polymerase chain reaction (PCR) was carried out using specific prim ers for ND4 and ND1 genes', 'Lab_Image: Capillary electrophoresis using an automated DNA sequencer to ana lyze PCR products', 'Lab_Image: Quantitative analysis of heteroplasmic mutation G11778A by prepar ing vector constructs and semiquantitative analyses', 'Lab_Image: PCR-restriction fragment length polymorphism (RFLP) was perform ed for T3394C and G11778A mutations', 'Lab_Image: MRI of mother: Mild atrophy of the optic nerve', 'Lab_Image: MRI of mother: Pons and cerebellum atrophy', 'Lab_Image: MRI of mother: No signal changes in the inferior olivary nucleus',

	'Lab_Image: Polymerase chain reaction (PCR) analysis of mtDNA: G11778A mutation', 'Lab_Image: Polymerase chain reaction (PCR) analysis of mtDNA: T3394C mutation', 'Lab_Image: Capillary electrophoresis using an automated DNA sequencer: Identification of G11778A and T3394C mutations', 'Lab_Image: PCR-restriction fragment length polymorphism (RFLP): HaeIII digestion for T3394C mutation, Tsp45I digestion for G11778A mutation', 'Lab_Image: Semiquantitative analysis of G11778A mutation: 92% heteroplasmic in III-1 and 70% heteroplasmic in II-2', 'Lab_Image: MRI: Atrophy of the optic nerve', 'Lab_Image: MRI: Severe atrophy of the cerebellum', 'Lab_Image: MRI: Mild atrophy of the brain stem', 'Lab_Image: MRI: Low signal intensities on T1-weighted imaging in bilateral inferior olivary nucleus (suggesting degeneration)', 'Lab_Image: MRI: High signal intensities on T2-weighted imaging in bilateral inferior olivary nucleus (suggesting degeneration)', 'Lab_Image: MRI: Atrophy of optic nerve and cerebellum, degenerative changes in bilateral inferior olivary nucleus', 'Lab_Image: MRI: Atrophy of the optic nerve', 'Lab_Image: MRI: Severe atrophy of the cerebellum', 'Lab_Image: MRI: Mild atrophy of the brain stem', 'Lab_Image: MRI: Low signal intensities on T1-weighted imaging in bilateral inferior olivary nucleus (suggesting degeneration)', 'Lab_Image: MRI: High signal intensities on T2-weighted imaging in bilateral inferior olivary nucleus (suggesting degeneration)', 'Lab_Image: PCR analysis: G11778A mutation identified', 'Lab_Image: PCR analysis: T3394C mutation identified', 'Lab_Image: Capillary electrophoresis using an automated DNA sequencer: G11778A and T3394C mutations detected', 'Lab_Image: Quantitative analysis of heteroplasmic mutation G11778A: III-1 - 92% heteroplasmic, II-2 - 70% heteroplasmic']}
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	'GI': [], 'GU': [], 'MSK': ['muscle: severe cerebellar ataxia', 'muscle: hypotonia', 'skeletal: atrophy of the optic nerve', 'skeletal: degenerative changes in the bilateral inferior olivary nucleus', 'skeletal: atrophy of the cerebellum and mild atrophy of the brain stem', 'skeletal: low signal intensities on T1-weighted imaging for the bilateral inferior olivary nucleus, suggesting degeneration'], 'ENDO': [], 'LYMPH': ['lymphatic_tissues: The bilateral inferior olivary nucleus exhibited low signal intensities on T1-weighted imaging, and high signal intensities on T2-weighted imaging, suggesting degeneration'], 'History': ['mutation: heteroplasmic G11778A and homoplasmic T3394C mutations in mitochondrial DNA', 'symptom: visual and gait disturbances that first appeared at 10 years of age', 'symptom: severe dizziness and double vision', 'total_iq: 73', 'performance_iq: 58', 'verbal_iq: 91', 'condition: bilateral exotropia, double vision', 'condition: incomplete horizontal movement of the eyes to the bilateral side', 'condition: horizontal and vertical gaze-evoked nystagmus', 'condition: dysarthria', 'condition: tremor in neck', 'condition: severe cerebellar ataxia and hypotonia', 'results: no abnormal findings', 'findings: no abnormal findings', 'condition: atrophy of optic nerve observed during ophthalmological examination', 'relative: mother (II-2)', 'condition: optic neuropathy', 'relative: uncle (II-3)', 'condition: optic neuropathy', 'relative: mother', 'condition: history of subarachnoid hemorrhage', 'results: mild atrophy of the optic nerve, pons, and cerebellum', 'symptom: visual and gait disturbances that first appeared at 10 years of age', 'symptom: severe dizziness and double vision', 'findings: bilateral exotropia, double vision', 'findings: incomplete horizontal movement of the eyes to the bilateral side', 'findings: horizontal and vertical gaze-evoked nystagmus', 'findings: dysarthria'], 'Lab_Image': ['Lab_Image: MRI disclosed atrophy of the optic nerve and cerebellum', 'Lab_Image: MRI disclosed degenerative changes in the bilateral inferior olivary nucleus', 'Lab_Image: orbital MRI revealed atrophy of the optic nerve', 'Lab_Image: brain MRI disclosed severe atrophy of the cerebellum and mild atrophy of the brain stem', 'Lab_Image: T1-weighted imaging showed low signal intensities on the bilateral inferior olivary nucleus', 'Lab_Image: T2-weighted imaging showed high signal intensities on the bilateral inferior olivary nucleus, suggesting degeneration',

	'Lab_Image: Blood samples were obtained from the patient and his mother with t heir informed consent', 'Lab_Image: The methods used for obtaining blood samples were approved by th e institutional review board of Tottori University Hospital', 'Lab_Image: mtDNA and genomic DNA extraction was carried out using standard procedures', 'Lab_Image: Polymerase chain reaction (PCR) was performed using specific prim ers for ND4 and ND1 genes', 'Lab_Image: Capillary electrophoresis using an automated DNA sequencer was u sed to analyze PCR products containing candidate abnormal points', 'Lab_Image: PCR-restriction fragment length polymorphism (RFLP) was carried out using HaeIII or Tsp45I enzymes to confirm mutations in mtDNA', 'Lab_Image: Semiquantitative analysis of G11778A mutation was performed usin g vector constructs and a mixture with several rate standards', 'Lab_Image: MRI disclosed atrophy of the optic nerve and cerebellum', 'Lab_Image: degenerative changes in the bilateral inferior olivary nucleus', 'Lab_Image: high signal intensities on T2-weighted imaging for the bilateral infer ior olivary nucleus, suggesting degeneration', 'Lab_Image: MRI of his mother disclosed mild atrophy of the optic nerve', 'Lab_Image: orbital MRI revealed atrophy of the optic nerve', 'Lab_Image: brain MRI disclosed severe atrophy of the cerebellum and mild atro phy of the brain stem']} [ ]:
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	'EENT': ['eyes: atrophy of the optic nerve', 'eyes: no pigmentation changes of the retina'], 'GI': [], 'GU': [], 'DERM': [], 'MSK': ['muscle: severe cerebellar ataxia', 'muscle: hypotonia', 'skeletal: atrophy of the optic nerve', 'skeletal: degenerative changes in the bilateral inferior olivary nucleus', 'skeletal: atrophy of the cerebellum and mild atrophy of the brain stem', 'skeletal: low signal intensities on T1-weighted imaging for the bilateral inferior olivary nucleus, suggesting degeneration'], 'ENDO': [], 'LYMPH': ['lymphatic_tissues: The bilateral inferior olivary nucleus exhibited low signal intensities on T1-weighted imaging, and high signal intensities on T2-weighted imaging, suggesting degeneration', 'lymphatic_tissues: Blood samples were obtained from the patient and his mother with their informed consent'], 'History': ['mutation: heteroplasmic G11778A and homoplasmic T3394C mutations in mitochondrial DNA', 'symptom: visual and gait disturbances that first appeared at 10 years of age', 'symptom: severe dizziness and double vision', 'total_iq: 73', 'performance_iq: 58', 'verbal_iq: 91', 'condition: bilateral exotropia, double vision', 'condition: incomplete horizontal movement of the eyes to the bilateral side', 'condition: horizontal and vertical gaze-evoked nystagmus', 'condition: dysarthria', 'condition: tremor in neck', 'condition: severe cerebellar ataxia and hypotonia', 'results: no abnormal findings', 'findings: no abnormal findings', 'condition: atrophy of optic nerve observed during ophthalmological examination', 'relative: mother (II-2)', 'condition: optic neuropathy', 'relative: uncle (II-3)', 'condition: optic neuropathy', 'relative: mother', 'condition: history of subarachnoid hemorrhage', 'results: mild atrophy of the optic nerve, pons, and cerebellum', 'symptom: visual and gait disturbances that first appeared at 10 years of age', 'symptom: severe dizziness and double vision', 'findings: bilateral exotropia, double vision', 'findings: incomplete horizontal movement of the eyes to the bilateral side', 'findings: horizontal and vertical gaze-evoked nystagmus', 'findings: dysarthria'], 'Lab_Image': ['Lab_Image: MRI disclosed atrophy of the optic nerve and cerebellum', 'Lab_Image: MRI disclosed degenerative changes in the bilateral inferior olivary nucleus', 'Lab_Image: orbital MRI revealed atrophy of the optic nerve', 'Lab_Image: brain MRI disclosed severe atrophy of the cerebellum and mild atrophy of the brain stem',

	'Lab_Image: T1-weighted imaging showed low signal intensities on the bilateral i nferior olivary nucleus', 'Lab_Image: T2-weighted imaging showed high signal intensities on the bilateral inferior olivary nucleus, suggesting degeneration', 'Lab_Image: Polymerase chain reaction (PCR) was carried out using specific prim ers for ND4 and ND1.', 'Lab_Image: Capillary electrophoresis using an automated DNA sequencer was u sed to analyze PCR products.', 'Lab_Image: Quantitative analysis of heteroplasmic mutation G11778A was perfo rmed using vector constructs and semiquantitative methods.', 'Lab_Image: PCR-restriction fragment length polymorphism (RFLP) was conduct ed using HaeIII for T3394C and Tsp45I for G11778A mutations.', 'Lab_Image: Blood samples were obtained from the patient and his mother with t heir informed consent', 'Lab_Image: The methods used were approved by the institutional review board of Tottori University Hospital', 'Lab_Image: mtDNA and genomic DNA were extracted by standard procedures', 'Lab_Image: Polymerase chain reaction (PCR) was carried out using specific prim ers for ND4 and ND1 genes', 'Lab_Image: PCR products were analyzed by capillary electrophoresis using an a utomated DNA sequencer', 'Lab_Image: The G11778A and T3394C mutations were identified in mtDNA', 'Lab_Image: Heteroplasmic mutation of G11778A was quantified using vector co nstructs and semiquantitative analysis', 'Lab_Image: Homoplasmic T3394C mutation was confirmed by PCR-restriction fr agment length polymorphism (RFLP)', 'Lab_Image: Differences between the patient and his mother were not observed f or the T3394C mutation', 'Lab_Image: Blood samples were obtained from the patient and his mother with t heir informed consent', 'Lab_Image: The methods used for obtaining blood samples were approved by th e institutional review board of Tottori University Hospital', 'Lab_Image: mtDNA and genomic DNA extraction was carried out using standard procedures', 'Lab_Image: Polymerase chain reaction (PCR) was conducted using specific prim ers for ND4 and ND1 genes', 'Lab_Image: Capillary electrophoresis using an automated DNA sequencer was u sed to analyze PCR products containing candidate abnormal points', 'Lab_Image: PCR-restriction fragment length polymorphism (RFLP) was perform ed on the PCR products, digesting them with HaeIII or Tsp45I for specific mutatio ns', 'Lab_Image: Semiquantitative analysis of G11778A mutation was conducted usin g a mixture of each vector construct with several rate standards', 'Lab_Image: MRI disclosed atrophy of the optic nerve and cerebellum', 'Lab_Image: degenerative changes in the bilateral inferior olivary nucleus', 'Lab_Image: high signal intensities on T2-weighted imaging for the bilateral infer ior olivary nucleus, suggesting degeneration', 'Lab_Image: polymerase chain reaction (PCR) using specific primers for ND4 and ND1 genes', 'Lab_Image: capillary electrophoresis using an automated DNA sequencer to anal yze PCR products', 'Lab_Image: PCR-restriction fragment length polymorphism (RFLP) using HaeIII or Tsp45I enzymes to confirm mtDNA mutations', 'Lab_Image: MRI of his mother disclosed mild atrophy of the optic nerve', 'Lab_Image: orbital MRI revealed atrophy of the optic nerve',

	'Lab_Image: brain MRI disclosed severe atrophy of the cerebellum and mild atrophy of the brain stem', 'Lab_Image: The methods used for extracting mtDNA and genomic DNA were approved by the institutional review board of Tottori University Hospital.', 'Lab_Image: Polymerase chain reaction (PCR) was carried out using specific primers for ND4 and ND1 genes to identify mutations related to LHON.', 'Lab_Image: PCR products were analyzed by capillary electrophoresis using an automated DNA sequencer.', 'Lab_Image: The presence of G11778A mutation in ND4 gene was confirmed through PCR-restriction fragment length polymorphism (RFLP) analysis.', 'Lab_Image: Heteroplasmic G11778A mutation levels were quantified using vector constructs and semiquantitative analyses.']} [ ]:
Qwen2:7B-Instruct Zero-shot prompts System-Wide Unified Extraction	{'Lab_Image': []}
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Qwen2.5:7B-Instruct	{'error': [], 'Lab_Image': []}

Few-Shot Prompt System-Wide Unified Extraction	
Qwen2.5:7B-Instruct Zero-Shot Prompts Category-Based Filtering and Targeted Extraction	{'iem': ['iem: is_IEM'], 'Vitals_Hema': [], 'Neuro': ['neurological: horizontal gaze palsy', 'neurological: gaze-evoked nystagmus', 'neurological: dysarthria', 'neurological: cerebellar ataxia', 'neurological: bilateral exotropia', 'neurological: double vision', 'neurological: incomplete horizontal movement of the eyes to the bilateral side', 'neurological: horizontal and vertical gaze-evoked nystagmus', 'neurological: tremor appeared in his neck', 'neurological: severe cerebellar ataxia and hypotonia in upper and lower limbs', 'neurological: no disturbances in cranial nerves I, VII, VIII, and XII were detected ', 'neurological: atrophy of the optic nerve', 'neurological: degeneration of the bilateral inferior olivary nucleus', 'cognitive: mild disturbance of cognitive function (Revised Wechsler Adult Intelli gence Scale: total IQ=73, performance IQ=58, verbal IQ=91)'], 'CVS': ['vascular: Atrophy of the optic nerve', 'vascular: Degeneration in the bilateral inferior olivary nucleus'], 'RESP': [], 'EENT': ['eyes: atrophy of the optic nerve', 'eyes: no pigmentation changes of the retina'], 'GI': [], 'GU': [], 'DERM': ['facial_features: bilateral exotropia', 'facial_features: double vision'], 'ENDO': [], 'LYMPH': [], 'History': ['past_medical_history: horizontal gaze palsy', 'past_medical_history: gaze-evoked nystagmus', 'past_medical_history: dysarthria', 'past_medical_history: cerebellar ataxia', 'past_medical_history: optic nerve and cerebellum atrophy', 'past_medical_history: degenerative changes in the bilateral inferior olivary nucl eus', 'history_of_present_illness: severe dizziness and double vision', 'history_of_present_illness: visual and gait disturbances first appeared at 10 year s of age', 'history_of_present_illness: bilateral exotropia, double vision, incomplete horizo ntal movement of the eyes to the bilateral side, horizontal, and vertical gaze-evok ed nystagmus, dysarthria', 'history_of_present_illness: mild disturbance of cognitive function (Revised Wec hsler Adult Intelligence Scale: total IQ=73, performance IQ=58, verbal IQ=91)', 'history_of_present_illness: neck tremor, no palatal myoclonus observed', 'history_of_present_illness: upper and lower limbs showed severe cerebellar ata xia and hypotonia', 'history_of_present_illness: no abnormal findings in deep tendon reflex and sens ory system',

	'history_of_present_illness: ophthalmological examination revealed optic nerve a trophy', 'family_and_genetics_history: mother (II-2) had optic neuropathy, history of sub arachnoid hemorrhage', 'family_and_genetics_history: uncle (II-3) had optic neuropathy', 'family_and_genetics_history: MRI of mother disclosed mild atrophy of the optic nerve, pons, and cerebellum', 'chief_complaint: severe dizziness and double vision'], 'Lab_Image': ['Lab_Image: Brain and orbit MRI disclosed atrophy of the optic nerv e and cerebellum, and degenerative changes in the bilateral inferior olivary nucle us', 'Lab_Image: Orbital MRI revealed atrophy of the optic nerve (Fig. 2A)', 'Lab_Image: Brain MRI disclosed severe atrophy of the cerebellum and mild atro phy of the brain stem (Fig. 2B)', 'Lab_Image: The bilateral inferior olivary nucleus exhibited low signal intensities on T1-weighted imaging, and high signal intensities on T2-weighted imaging, sug gesting degeneration (Fig. 2C and D)', 'Lab_Image: Orbital MRI revealed atrophy of the optic nerve (Fig. 2A).', 'Lab_Image: Brain MRI disclosed severe atrophy of the cerebellum and mild atro phy of the brain stem (Fig. 2B).', 'Lab_Image: The bilateral inferior olivary nucleus exhibited low signal intensities on T1-weighted imaging, and high signal intensities on T2-weighted imaging, sug gesting degeneration (Fig. 2C and D).', 'Lab_Image: MRI of his mother disclosed mild atrophy of the optic nerve (Fig. 2 E), pons, and cerebellum (Fig. 2F-H).', 'Lab_Image: Ophthalmological examination revealed atrophy of the optic nerve, but there were no pigmentation changes of the retina.', 'Lab_Image: Orbital MRI revealed atrophy of the optic nerve (Fig. 2A).', 'Lab_Image: Brain MRI disclosed severe atrophy of the cerebellum and mild atro phy of the brain stem (Fig. 2B).', 'Lab_Image: The bilateral inferior olivary nucleus exhibited low signal intensities on T1-weighted imaging, and high signal intensities on T2-weighted imaging, sug gesting degeneration (Fig. 2C and D).', 'Lab_Image: MRI of his mother disclosed mild atrophy of the optic nerve (Fig. 2 E), pons, and cerebellum (Fig. 2F-H).', 'Lab_Image: Orbital MRI revealed atrophy of the optic nerve.', 'Lab_Image: Brain MRI disclosed severe atrophy of the cerebellum and mild atro phy of the brain stem.', 'Lab_Image: The bilateral inferior olivary nucleus exhibited low signal intensities on T1-weighted imaging, and high signal intensities on T2-weighted imaging, sug gesting degeneration.']} [ ]:
Qwen2.5:7B-Instruct Zero-shot prompts Uniform Application of Category Prompts	{'iem': ['iem: is_IEM'], 'Vitals_Hema': [], 'Pregnancy': ['maternal_health: The T3394C mutation present in both the proban d (III-1) and his mother (II-2) was homoplasmic; differences between the patient and his mother were not observed for this mutation.', 'maternal_health: The G11778A mutation, which causes an Arg-to-His amino aci d substitution in ND4, was also observed.'], 'Neuro': ['neurological: horizontal gaze palsy', 'neurological: gaze-evoked nystagmus', 'neurological: dysarthria',

	'neurological: cerebellar ataxia', 'neurological: bilateral exotropia', 'neurological: double vision', 'neurological: incomplete horizontal movement of the eyes to the bilateral side', 'neurological: horizontal and vertical gaze-evoked nystagmus', 'neurological: tremor appeared in his neck', 'neurological: severe cerebellar ataxia and hypotonia in upper and lower limbs', 'neurological: no disturbances in cranial nerves I, VII, VIII, and XII were detected ', 'neurological: atrophy of the optic nerve', 'neurological: degeneration of the bilateral inferior olivary nucleus', 'cognitive: mild disturbance of cognitive function (Revised Wechsler Adult Intelli gence Scale: total IQ=73, performance IQ=58, verbal IQ=91)'], 'CVS': ['vascular: Atrophy of the optic nerve', 'vascular: Degeneration in the bilateral inferior olivary nucleus'], 'RESP': [], 'EENT': ['eyes: atrophy of the optic nerve', 'eyes: no pigmentation changes of the retina'], 'GI': [], 'GU': [], 'DERM': ['facial_features: bilateral exotropia', 'facial_features: double vision'], 'MSK': [], 'ENDO': [], 'LYMPH': [], 'History': ['past_medical_history: horizontal gaze palsy', 'past_medical_history: gaze-evoked nystagmus', 'past_medical_history: dysarthria', 'past_medical_history: cerebellar ataxia', 'past_medical_history: optic nerve and cerebellum atrophy', 'past_medical_history: degenerative changes in the bilateral inferior olivary nucl eus', 'history_of_present_illness: severe dizziness and double vision', 'history_of_present_illness: visual and gait disturbances first appeared at 10 year s of age', 'history_of_present_illness: bilateral exotropia, double vision, incomplete horizo ntal movement of the eyes to the bilateral side, horizontal, and vertical gaze-evok ed nystagmus, dysarthria', 'history_of_present_illness: mild disturbance of cognitive function (Revised Wec hsler Adult Intelligence Scale: total IQ=73, performance IQ=58, verbal IQ=91)', 'history_of_present_illness: neck tremor, no palatal myoclonus observed', 'history_of_present_illness: upper and lower limbs showed severe cerebellar ata xia and hypotonia', 'history_of_present_illness: no abnormal findings in deep tendon reflex and sens ory system', 'history_of_present_illness: ophthalmological examination revealed optic nerve a trophy', 'family_and_genetics_history: mother (II-2) had optic neuropathy, history of sub arachnoid hemorrhage', 'family_and_genetics_history: uncle (II-3) had optic neuropathy', 'family_and_genetics_history: MRI of mother disclosed mild atrophy of the optic nerve, pons, and cerebellum', 'chief_complaint: severe dizziness and double vision',

	'family_and_genetics_history: The patient and his mother were found to have the homoplasmic T3394C mutation in mtDNA, which causes a Tyr-to-His amino acid substitution in ND1.', 'family_and_genetics_history: Both the proband (III-1) and his mother (II-2) had heteroplasmic G11778A mutations in mtDNA, with III-1 having 92% and II-2 having 70% heteroplasmy. This mutation causes an Arg-to-His amino acid substitution in ND4.'], 'Lab_Image': ['Lab_Image: Blood samples were obtained from the patient and his mother with their informed consent.', 'Lab_Image: mtDNA and genomic DNA were extracted by standard procedures.', 'Lab_Image: The polymerase chain reaction (PCR) was carried out using specific primers for ND4 and ND1 to detect mutations.', 'Lab_Image: PCR products that included the previously reported candidate abnormal points were analyzed by capillary electrophoresis using an automated DNA sequencer.', 'Lab_Image: The G11778A and T3394C mutations were identified, while the A3243G mutation was not detected.', 'Lab_Image: The mutations in the mtDNA were confirmed by performing PCR-restriction fragment length polymorphism (RFLP), in which the PCR products were digested using either HaeIII (for T3394C) or Tsp45I (for G11778A).', 'Lab_Image: Semiquantitative analyses of G11778A were performed using a mixture of each with several rate standards.', 'Lab_Image: Sequence analysis revealed the homoplasmic T3394C mutation of the mtDNA.', 'Lab_Image: PCR-RFLP data showed that the T3394C mutation present in both the proband (III-1) and his mother (II-2) was homoplasmic; differences between the patient and his mother were not observed for this mutation.', 'Lab_Image: The G11778A mutation, which causes an Arg-to-His amino acid substitution in ND4, was also observed.', 'Lab_Image: PCR-RFLP data showed that this mutation in the patient and his mother was heteroplasmic.', 'Lab_Image: The semiquantitative analysis performed to determine the effect of this mutation on disease severity revealed that III-1 had a 92% heteroplasmic G11778A mutation, and II-2 had a 70% heteroplasmic G11778A mutation.', 'Lab_Image: Brain and orbit MRI disclosed atrophy of the optic nerve and cerebellum, and degenerative changes in the bilateral inferior olivary nucleus', 'Lab_Image: Orbital MRI revealed atrophy of the optic nerve (Fig. 2A)', 'Lab_Image: Brain MRI disclosed severe atrophy of the cerebellum and mild atrophy of the brain stem (Fig. 2B)', 'Lab_Image: The bilateral inferior olivary nucleus exhibited low signal intensities on T1-weighted imaging, and high signal intensities on T2-weighted imaging, suggesting degeneration (Fig. 2C and D)', 'Lab_Image: Orbital MRI revealed atrophy of the optic nerve (Fig. 2A).', 'Lab_Image: Brain MRI disclosed severe atrophy of the cerebellum and mild atrophy of the brain stem (Fig. 2B).', 'Lab_Image: The bilateral inferior olivary nucleus exhibited low signal intensities on T1-weighted imaging, and high signal intensities on T2-weighted imaging, suggesting degeneration (Fig. 2C and D).', 'Lab_Image: MRI of his mother disclosed mild atrophy of the optic nerve (Fig. 2E), pons, and cerebellum (Fig. 2F-H).', 'Lab_Image: PCR was carried out using primers for ND4 and ND1 to detect mutations in mtDNA.', 'Lab_Image: Capillary electrophoresis using an automated DNA sequencer was performed on PCR products that included the previously reported candidate abnormal points.',

	'Lab_Image: PCR-RFLP data revealed a homoplasmic T3394C mutation present i n both the proband (III-1) and his mother (II-2).', 'Lab_Image: The G11778A mutation, which causes an Arg-to-His amino acid subs titution in ND4, was also observed.', 'Lab_Image: PCR-RFLP data showed that this mutation in the patient and his mot her was heteroplasmic.', 'Lab_Image: Semiquantitative analysis performed to determine the effect of the G11778A mutation on disease severity revealed that III-1 had a 92% heteroplasm ic G11778A mutation, and II-2 had a 70% heteroplasmic G11778A mutation.', 'Lab_Image: Ophthalmological examination revealed atrophy of the optic nerve, but there were no pigmentation changes of the retina.', 'Lab_Image: Orbital MRI revealed atrophy of the optic nerve (Fig. 2A).', 'Lab_Image: Brain MRI disclosed severe atrophy of the cerebellum and mild atro phy of the brain stem (Fig. 2B).', 'Lab_Image: The bilateral inferior olivary nucleus exhibited low signal intensities on T1-weighted imaging, and high signal intensities on T2-weighted imaging, sug gesting degeneration (Fig. 2C and D).', 'Lab_Image: MRI of his mother disclosed mild atrophy of the optic nerve (Fig. 2 E), pons, and cerebellum (Fig. 2F-H).', 'Lab_Image: Orbital MRI revealed atrophy of the optic nerve.', 'Lab_Image: Brain MRI disclosed severe atrophy of the cerebellum and mild atro phy of the brain stem.', 'Lab_Image: The bilateral inferior olivary nucleus exhibited low signal intensities on T1-weighted imaging, and high signal intensities on T2-weighted imaging, sug gesting degeneration.']}
Qwen2.5:7B-Instruct Zero-shot prompts System-Wide Unified Extraction	{'iem': ['iem: is_IEM'], 'Vitals_Hema': ['Vitals_Hema: temperature', 'Vitals_Hema: pulse', 'Vitals_Hema: respiratory_rate', 'Vitals_Hema: blood_pressure', 'Vitals_Hema: oxygen_saturation', 'Vitals_Hema: hematological_conditions', 'Vitals_Hema: hematology_lab_tests_measurements'], 'Pregnancy': ['Pregnancy: neonatal_health', 'Pregnancy: maternal_health', 'Pregnancy: pregnancy_lab_tests_imaging_exam'], 'Neuro': ['Neuro: neurological', 'Neuro: cognitive', 'Neuro: neuro_lab_tests_imaging_exam'], 'EENT': ['EENT: eyes', 'EENT: ears', 'EENT: nose', 'EENT: throat', 'EENT: EENT_lab_tests_imaging_exam'], 'CVS': ['CVS: cardiac', 'CVS: vascular', 'CVS: CVS_lab_tests_imaging_exam'], 'RESP': ['RESP: respiratory', 'RESP: respiratory_system_lab_tests_imaging_exam'], 'GI': ['GI: gastrointestinal', 'GI: gastrointestinal_lab_tests_imaging_exam'], 'GU': ['GU: genital', 'GU: urinary', 'GU: GU_lab_tests_imaging_exam'], 'DERM': ['DERM: skin_conditions', 'DERM: facial_features', 'DERM: breast_conditions', 'DERM: derm_breast_facial_lab_tests_image_exam'], 'MSK': ['MSK: muscle', 'MSK: skeletal', 'MSK: MSK_lab_tests_image_exam'],

	'LYMPH': ['LYMPH: adenoid', 'LYMPH: tonsils', 'LYMPH: lymphatic_tissues', 'LYMPH: lymph_nodes', 'LYMPH: bone_marrow', 'LYMPH: spleen', 'LYMPH: immune_cells', 'LYMPH: Lymphatic_lab_tests_image_exam'], 'ENDO': ['ENDO: endocrine_glands', 'ENDO: Endocrine_lab_tests_image_exam'], 'History': ['History: past_medical_history', 'History: past_surgical_history', 'History: history_of_present_illness', 'History: social_history', 'History: family_and_genetics_history', 'History: chief_complaint'], 'Lab_Image': []}
Qwen2.5:32B-Instruct Few-Shot Prompts Category-Based Filtering and Targeted Extraction	{'iem': ['iem: Inborn errors of metabolism'], 'Vitals_Hema': [], 'Pregnancy': ['maternal_health: History of subarachnoid hemorrhage', 'maternal_health: Mild atrophy of the optic nerve', 'maternal_health: Pons, and cerebellum'], 'Neuro': ['neurological: horizontal gaze palsy', 'neurological: gaze-evoked nystagmus', 'neurological: dysarthria', 'neurological: cerebellar ataxia', 'neurological: bilateral exotropia, double vision, incomplete horizontal movemen t of the eyes to the bilateral side, horizontal and vertical gaze-evoked nystagmus, dysarthria', 'neurological: tremor appeared in his neck', 'neurological: severe cerebellar ataxia and hypotonia', 'neurological: atrophy of the optic nerve', 'neurological: severe atrophy of the cerebellum and mild atrophy of the brain ste m', 'neurological: bilateral inferior olivary nucleus exhibited low signal intensities o n T1-weighted imaging, and high signal intensities on T2-weighted imaging, sugg esting degeneration', 'cognitive: mild disturbance of cognitive function (Revised Wechsler Adult Intelli gence Scale: total IQ=73, performance IQ=58, verbal IQ=91)'], 'RESP': [], 'EENT': ['eyes: bilateral exotropia', 'eyes: double vision', 'eyes: incomplete horizontal movement of the eyes to the bilateral side', 'eyes: horizontal, and vertical gaze-evoked nystagmus', 'throat: dysarthria'], 'GI': [], 'GU': [], 'DERM': ['facial_features: bilateral exotropia', 'facial_features: double vision', 'facial_features: incomplete horizontal movement of the eyes to the bilateral side '], 'MSK': ['muscle: his upper and lower limbs showed no paralysis, they exhibited s evere cerebellar ataxia and hypotonia'],

	'ENDO': ['endocrine_glands: Although the thyrotropin-releasing drug taltirelin did not relieve his symptoms'], 'LYMPH': [], 'Lab_Image': ['Lab_Image: Blood and cerebrospinal fluid analyses were normal', 'Lab_Image: MRI of his mother disclosed mild atrophy of the optic nerve', 'Lab_Image: pons, and cerebellum', 'Lab_Image: Brain and orbit MRI disclosed atrophy of the optic nerve and cerebellum, and degenerative changes in the bilateral inferior olivary nucleus', 'Lab_Image: Ophthalmological examination revealed atrophy of the optic nerve', 'Lab_Image: Orbital MRI revealed atrophy of the optic nerve', 'Lab_Image: brain MRI disclosed severe atrophy of the cerebellum and mild atrophy of the brain stem', 'Lab_Image: MRI of his mother disclosed mild atrophy of the optic nerve, pons, and cerebellum', 'Lab_Image: Ophthalmological examination revealed atrophy of the optic nerve, but there were no pigmentation changes of the retina.', 'Lab_Image: Orbital MRI revealed atrophy of the optic nerve', 'Lab_Image: MRI of his mother disclosed mild atrophy of the optic nerve']} [ ]:
Qwen2.5:32B-Instruct Few-shot prompts Uniform Application of Category Prompts	{'iem': ['iem: Inborn errors of metabolism'], 'Vitals_Hema': ['hematological_conditions: T3394C mutation of the mtDNA', 'hematological_conditions: G11778A mutation in ND4'], 'Pregnancy': ['maternal_health: History of subarachnoid hemorrhage', 'maternal_health: Mild atrophy of the optic nerve', 'maternal_health: Pons, and cerebellum'], 'Neuro': ['neurological: horizontal gaze palsy', 'neurological: gaze-evoked nystagmus', 'neurological: dysarthria', 'neurological: cerebellar ataxia', 'neurological: bilateral exotropia, double vision, incomplete horizontal movement of the eyes to the bilateral side, horizontal and vertical gaze-evoked nystagmus, dysarthria', 'neurological: tremor appeared in his neck', 'neurological: severe cerebellar ataxia and hypotonia', 'neurological: atrophy of the optic nerve', 'neurological: severe atrophy of the cerebellum and mild atrophy of the brain stem', 'neurological: bilateral inferior olivary nucleus exhibited low signal intensities on T1-weighted imaging, and high signal intensities on T2-weighted imaging, suggesting degeneration', 'cognitive: mild disturbance of cognitive function (Revised Wechsler Adult Intelligence Scale: total IQ=73, performance IQ=58, verbal IQ=91)', 'neurological: Established genetic abnormalities associated with cerebellar ataxia including polyglutamine diseases were not found.'], 'CVS': ['cardiac: This mutation causes a Tyr-to-His amino acid substitution in ND1', 'cardiac: The G11778A mutation, which causes an Arg-to-His amino acid substitution in ND4'], 'RESP': [], 'EENT': ['eyes: bilateral exotropia', 'eyes: double vision', 'eyes: incomplete horizontal movement of the eyes to the bilateral side',

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Qwen2.5:32B-Instruct Few-Shot Prompt	{'error': [], 'Lab_Image': []}

System-Wide Unified Extraction	
Qwen2.5:32B-Instruct Zero-Shot Prompts Category-Based Filtering and Targeted Extraction	{'iem': ['iem: is_IEM'], 'Vitals_Hema': [], 'Neuro': ['neurological: horizontal gaze palsy', 'neurological: gaze-evoked nystagmus', 'neurological: dysarthria', 'neurological: cerebellar ataxia', 'neurological: bilateral exotropia, double vision, incomplete horizontal movemen t of the eyes to the bilateral side, horizontal and vertical gaze-evoked nystagmus, dysarthria', 'neurological: atrophy of the optic nerve', 'neurological: severe cerebellar ataxia and hypotonia in upper and lower limbs', 'neurological: degeneration in the bilateral inferior olivary nucleus', 'cognitive: mild disturbance of cognitive function (Revised Wechsler Adult Intelli gence Scale: total IQ=73, performance IQ=58, verbal IQ=91)'], 'RESP': [], 'EENT': ['eyes: bilateral exotropia', 'eyes: double vision', 'eyes: incomplete horizontal movement of the eyes to the bilateral side', 'eyes: horizontal gaze-evoked nystagmus', 'eyes: vertical gaze-evoked nystagmus'], 'GI': [], 'GU': [], 'DERM': ['facial_features: bilateral exotropia', 'facial_features: double vision', 'facial_features: incomplete horizontal movement of the eyes to the bilateral side '], 'MSK': ['muscle: his upper and lower limbs showed no paralysis, they exhibited s evere cerebellar ataxia and hypotonia.'], 'ENDO': ['endocrine_glands: Although the thyrotropin-releasing drug taltirelin di d not relieve his symptoms'], 'LYMPH': [], 'History': ['past_medical_history: visual and gait disturbances first appeared at 10 years of age', 'past_medical_history: horizontal gaze palsy, gaze-evoked nystagmus, dysarthria, cerebellar ataxia', 'past_medical_history: atrophy of the optic nerve and cerebellum, degenerative c hanges in the bilateral inferior olivary nucleus', 'history_of_present_illness: severe dizziness and double vision', 'history_of_present_illness: mild disturbance of cognitive function (Revised Wec hsler Adult Intelligence Scale: total IQ=73, performance IQ=58, verbal IQ=91)', 'history_of_present_illness: bilateral exotropia, double vision, incomplete horizo ntal movement of the eyes to the bilateral side, horizontal and vertical gaze-evoke d nystagmus, dysarthria', 'history_of_present_illness: severe cerebellar ataxia and hypotonia in upper and l ower limbs', 'family_and_genetics_history: mother (II-2) and uncle (II-3) also had optic neuro pathy', 'family_and_genetics_history: mother has a history of subarachnoid hemorrhage ', 'family_and_genetics_history: MRI of his mother disclosed mild atrophy of the op tic nerve, pons, and cerebellum', 'chief_complaint: severe dizziness and double vision'],

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Qwen2.5:32B-Instruct Zero-shot prompts Uniform Application of Category Prompts	{'iem': ['iem: is_IEM'], 'Vitals_Hema': ['hematological_conditions: mitochondrial encephalomyopathy, lactic acidosis, and stroke-like episodes'], 'Pregnancy': [], 'Neuro': ['neurological: horizontal gaze palsy', 'neurological: gaze-evoked nystagmus', 'neurological: dysarthria', 'neurological: cerebellar ataxia', 'neurological: bilateral exotropia, double vision, incomplete horizontal movement of the eyes to the bilateral side, horizontal and vertical gaze-evoked nystagmus, dysarthria', 'neurological: atrophy of the optic nerve', 'neurological: severe cerebellar ataxia and hypotonia in upper and lower limbs', 'neurological: degeneration in the bilateral inferior olivary nucleus', 'cognitive: mild disturbance of cognitive function (Revised Wechsler Adult Intelligence Scale: total IQ=73, performance IQ=58, verbal IQ=91)', 'neurological: The G11778A mutation, which causes an Arg-to-His amino acid substitution in ND4', 'neurological: The T3394C mutation causes a Tyr-to-His amino acid substitution in ND1'], 'CVS': ['cardiac: mitochondrial encephalomyopathy, lactic acidosis, and stroke-like episodes'], 'RESP': [], 'EENT': ['eyes: bilateral exotropia', 'eyes: double vision', 'eyes: incomplete horizontal movement of the eyes to the bilateral side', 'eyes: horizontal gaze-evoked nystagmus', 'eyes: vertical gaze-evoked nystagmus'], 'GI': [], 'GU': [], 'DERM': ['facial_features: bilateral exotropia', 'facial_features: double vision', 'facial_features: incomplete horizontal movement of the eyes to the bilateral side'], 'MSK': ['muscle: his upper and lower limbs showed no paralysis, they exhibited severe cerebellar ataxia and hypotonia.'], 'ENDO': ['endocrine_glands: Although the thyrotropin-releasing drug taltirelin did not relieve his symptoms'], 'LYMPH': [],

	'History': ['past_medical_history: visual and gait disturbances first appeared at 10 years of age', 'past_medical_history: horizontal gaze palsy, gaze-evoked nystagmus, dysarthria, cerebellar ataxia', 'past_medical_history: atrophy of the optic nerve and cerebellum, degenerative changes in the bilateral inferior olivary nucleus', 'history_of_present_illness: severe dizziness and double vision', 'history_of_present_illness: mild disturbance of cognitive function (Revised Wechsler Adult Intelligence Scale: total IQ=73, performance IQ=58, verbal IQ=91)', 'history_of_present_illness: bilateral exotropia, double vision, incomplete horizontal movement of the eyes to the bilateral side, horizontal and vertical gaze-evoked nystagmus, dysarthria', 'history_of_present_illness: severe cerebellar ataxia and hypotonia in upper and lower limbs', 'family_and_genetics_history: mother (II-2) and uncle (II-3) also had optic neuropathy', 'family_and_genetics_history: mother has a history of subarachnoid hemorrhage', 'family_and_genetics_history: MRI of his mother disclosed mild atrophy of the optic nerve, pons, and cerebellum', 'chief_complaint: severe dizziness and double vision', 'family_and_genetics_history: The G11778A mutation, which causes an Arg-to-His amino acid substitution in ND4, was also observed.', 'family_and_genetics_history: PCR-RFLP data showed that this mutation in the patient and his mother was heteroplasmic.', 'family_and_genetics_history: The semiquantitative analysis performed to determine the effect of this mutation on disease severity revealed that III-1 had a 92% heteroplasmic G11778A mutation, and II-2 had a 70% heteroplasmic G11778A mutation.', 'family_and_genetics_history: The mutations in mtDNA include homoplasmic T3394C mutation of the mtDNA which causes a Tyr-to-His amino acid substitution in ND1.'], 'Lab_Image': ['Lab_Image: G11778A mutation heteroplasmic levels: III-1 (92%), II-2 (70%)', 'Lab_Image: homoplasmic T3394C mutation of the mtDNA', 'Lab_Image: The G11778A mutation, which causes an Arg-to-His amino acid substitution in ND4, was also observed.', 'Lab_Image: PCR-RFLP data showed that this mutation in the patient and his mother was heteroplasmic.', 'Lab_Image: Semiquantitative analysis performed to determine the effect of this mutation on disease severity revealed that III-1 had a 92% heteroplasmic G11778A mutation, and II-2 had a 70% heteroplasmic G11778A mutation.', 'Lab_Image: Sequence analysis revealed the homoplasmic T3394C mutation of the mtDNA.', 'Lab_Image: This mutation causes a Tyr-to-His amino acid substitution in ND1.', 'Lab_Image: Brain and orbit MRI disclosed atrophy of the optic nerve and cerebellum, and degenerative changes in the bilateral inferior olivary nucleus', 'Lab_Image: Ophthalmological examination revealed atrophy of the optic nerve', 'Lab_Image: Orbital MRI revealed atrophy of the optic nerve', 'Lab_Image: brain MRI disclosed severe atrophy of the cerebellum and mild atrophy of the brain stem', 'Lab_Image: The bilateral inferior olivary nucleus exhibited low signal intensities on T1-weighted imaging, and high signal intensities on T2-weighted imaging, suggesting degeneration',

	'Lab_Image: PCR products that included the previously reported candidate abnormal points were analyzed by capillary electrophoresis using an automated DNA sequencer.', 'Lab_Image: The G11778A and T3394C mutations were identified, while the A3243G mutation was not detected.', 'Lab_Image: Sequence analysis revealed the homoplasmic T3394C mutation of the mtDNA', 'Lab_Image: PCR-RFLP data revealed that the T3394C mutation present in both the proband (III-1) and his mother (II-2) was homoplasmic; differences between the patient and his mother were not observed for this mutation.', 'Lab_Image: The G11778A mutation, which causes an Arg-to-His amino acid substitution in ND4, was also observed', 'Lab_Image: PCR-RFLP data showed that this mutation in the patient and his mother was heteroplasmic', 'Lab_Image: Semiquantitative analysis performed to determine the effect of this mutation on disease severity revealed that III-1 had a 92% heteroplasmic G11778A mutation, and II-2 had a 70% heteroplasmic G11778A mutation', 'Lab_Image: PCR products that included the previously reported candidate abnormal points were analyzed by capillary electrophoresis using an automated DNA sequencer.', 'Lab_Image: The G11778A and T3394C mutations were identified, while the A3243G mutation was not detected.', 'Lab_Image: Sequence analysis revealed the homoplasmic T3394C mutation of the mtDNA.', 'Lab_Image: PCR-RFLP data revealed that the T3394C mutation present in both the proband (III-1) and his mother (II-2) was homoplasmic; differences between the patient and his mother were not observed for this mutation.', 'Lab_Image: The G11778A mutation, which causes an Arg-to-His amino acid substitution in ND4, was also observed.', 'Lab_Image: PCR-RFLP data showed that this mutation in the patient and his mother was heteroplasmic.', 'Lab_Image: Semiquantitative analysis performed to determine the effect of this mutation on disease severity revealed that III-1 had a 92% heteroplasmic G11778A mutation, and II-2 had a 70% heteroplasmic G11778A mutation.', 'Lab_Image: Ophthalmological examination revealed atrophy of the optic nerve, but there were no pigmentation changes of the retina.', 'Lab_Image: Orbital MRI revealed atrophy of the optic nerve.', 'Lab_Image: MRI of his mother disclosed mild atrophy of the optic nerve.']}
Qwen2.5:32B-Instruct Zero-shot prompts System-Wide Unified Extraction	'iem': ['iem: is_IEM'], 'Vitals_Hema': ['Vitals_Hema: temperature', 'Vitals_Hema: pulse', 'Vitals_Hema: respiratory_rate', 'Vitals_Hema: blood_pressure', 'Vitals_Hema: oxygen_saturation', 'Vitals_Hema: hematological_conditions', 'Vitals_Hema: hematology_lab_tests_measurements'], 'Pregnancy': ['Pregnancy: neonatal_health', 'Pregnancy: maternal_health', 'Pregnancy: pregnancy_lab_tests_imaging_exam'], 'Neuro': ['Neuro: neurological', 'Neuro: cognitive', 'Neuro: neuro_lab_tests_imaging_exam'], 'EENT': ['EENT: eyes',

	'EENT: ears', 'EENT: nose', 'EENT: throat', 'EENT: EENT_lab_tests_imaging_exam'], 'CVS': ['CVS: cardiac', 'CVS: vascular', 'CVS: CVS_lab_tests_imaging_exam'], 'RESP': ['RESP: respiratory', 'RESP: respiratory_system_lab_tests_imaging_exam'], 'GI': ['GI: gastrointestinal', 'GI: gastrointestinal_lab_tests_imaging_exam'], 'GU': ['GU: genital', 'GU: urinary', 'GU: GU_lab_tests_imaging_exam'], 'DERM': ['DERM: skin_conditions', 'DERM: facial_features', 'DERM: breast_conditions', 'DERM: derm_breast_facial_lab_tests_image_exam'], 'MSK': ['MSK: muscle', 'MSK: skeletal', 'MSK: MSK_lab_tests_image_exam'], 'LYMPH': ['LYMPH: adenoid', 'LYMPH: tonsils', 'LYMPH: lymphatic_tissues', 'LYMPH: lymph_nodes', 'LYMPH: bone_marrow', 'LYMPH: spleen', 'LYMPH: immune_cells', 'LYMPH: Lymphatic_lab_tests_image_exam'], 'ENDO': ['ENDO: endocrine_glands', 'ENDO: Endocrine_lab_tests_image_exam'], 'History': ['History: past_medical_history', 'History: past_surgical_history', 'History: history_of_present_illness', 'History: social_history', 'History: family_and_genetics_history', 'History: chief_complaint'], 'Lab_Image': []}
Qwen2.5:32B-Instruct Zero-Shot Prompts Chain-of-Thought Category-Based Filtering and Targeted Extraction	{'iem': ['iem: is_IEM'], 'Vitals_Hema': [], 'Neuro': ['neurological: horizontal gaze palsy', 'neurological: gaze-evoked nystagmus', 'neurological: dysarthria', 'neurological: cerebellar ataxia', 'neurological: bilateral exotropia, double vision, incomplete horizontal movemen t of the eyes to the bilateral side, horizontal and vertical gaze-evoked nystagmus, dysarthria', 'neurological: atrophy of the optic nerve', 'neurological: severe cerebellar ataxia and hypotonia in upper and lower limbs', 'neurological: degeneration in the bilateral inferior olivary nucleus', 'cognitive: mild disturbance of cognitive function (Revised Wechsler Adult Intelli gence Scale: total IQ=73, performance IQ=58, verbal IQ=91)'], 'RESP': [], 'EENT': ['eyes: bilateral exotropia', 'eyes: double vision', 'eyes: incomplete horizontal movement of the eyes to the bilateral side', 'eyes: horizontal gaze-evoked nystagmus', 'eyes: vertical gaze-evoked nystagmus'], 'GI': [], 'GU': [], 'DERM': ['facial_features: bilateral exotropia', 'facial_features: double vision',

	'facial_features: incomplete horizontal movement of the eyes to the bilateral side '], 'MSK': ['muscle: his upper and lower limbs showed no paralysis, they exhibited s evere cerebellar ataxia and hypotonia.'], 'ENDO': ['endocrine_glands: Although the thyrotropin-releasing drug taltirelin di d not relieve his symptoms'], 'LYMPH': [], 'History': ['past_medical_history: visual and gait disturbances first appeared at 10 years of age', 'past_medical_history: horizontal gaze palsy, gaze-evoked nystagmus, dysarthria, cerebellar ataxia', 'past_medical_history: atrophy of the optic nerve and cerebellum, degenerative c hanges in the bilateral inferior olivary nucleus', 'history_of_present_illness: severe dizziness and double vision', 'history_of_present_illness: mild disturbance of cognitive function (Revised Wec hsler Adult Intelligence Scale: total IQ=73, performance IQ=58, verbal IQ=91)', 'history_of_present_illness: bilateral exotropia, double vision, incomplete horizo ntal movement of the eyes to the bilateral side, horizontal and vertical gaze-evoke d nystagmus, dysarthria', 'history_of_present_illness: severe cerebellar ataxia and hypotonia in upper and l ower limbs', 'family_and_genetics_history: mother (II-2) and uncle (II-3) also had optic neuro pathy', 'family_and_genetics_history: mother has a history of subarachnoid hemorrhage ', 'family_and_genetics_history: MRI of his mother disclosed mild atrophy of the op tic nerve, pons, and cerebellum', 'chief_complaint: severe dizziness and double vision'], 'Lab_Image': ['Lab_Image: Brain and orbit MRI disclosed atrophy of the optic nerv e and cerebellum, and degenerative changes in the bilateral inferior olivary nucle us', 'Lab_Image: Ophthalmological examination revealed atrophy of the optic nerve', 'Lab_Image: Orbital MRI revealed atrophy of the optic nerve', 'Lab_Image: brain MRI disclosed severe atrophy of the cerebellum and mild atro phy of the brain stem', 'Lab_Image: The bilateral inferior olivary nucleus exhibited low signal intensities on T1-weighted imaging, and high signal intensities on T2-weighted imaging, sug gesting degeneration', 'Lab_Image: Ophthalmological examination revealed atrophy of the optic nerve, but there were no pigmentation changes of the retina.', 'Lab_Image: Orbital MRI revealed atrophy of the optic nerve.', 'Lab_Image: MRI of his mother disclosed mild atrophy of the optic nerve.']}
Qwen2.5:32B-Instruct Zero-shot prompts Chain-of-Thought Uniform Application of Category Prompts	{'iem': ['iem: is_IEM'], 'Vitals_Hema': ['hematological_conditions: mitochondrial encephalomyopathy, la ctic acidosis, and stroke-like episodes'], 'Pregnancy': [], 'Neuro': ['neurological: horizontal gaze palsy', 'neurological: gaze-evoked nystagmus', 'neurological: dysarthria', 'neurological: cerebellar ataxia', 'neurological: bilateral exotropia, double vision, incomplete horizontal movemen t of the eyes to the bilateral side, horizontal and vertical gaze-evoked nystagmus, dysarthria', 'neurological: atrophy of the optic nerve',

	'neurological: severe cerebellar ataxia and hypotonia', 'neurological: degeneration in the bilateral inferior olivary nucleus', 'cognitive: mild disturbance of cognitive function (Revised Wechsler Adult Intelligence Scale: total IQ=73, performance IQ=58, verbal IQ=91)', 'neurological: The G11778A mutation, which causes an Arg-to-His amino acid substitution in ND4', 'neurological: The T3394C mutation causes a Tyr-to-His amino acid substitution in ND1'], 'CVS': ['cardiac: mitochondrial encephalomyopathy, lactic acidosis, and stroke-like episodes'], 'RESP': [], 'EENT': ['eyes: bilateral exotropia', 'eyes: double vision', 'eyes: incomplete horizontal movement of the eyes to the bilateral side', 'eyes: horizontal gaze-evoked nystagmus', 'eyes: vertical gaze-evoked nystagmus', 'eyes: atrophy of the optic nerve', 'throat: dysarthria'], 'GI': [], 'GU': [], 'DERM': ['facial_features: bilateral exotropia', 'facial_features: double vision', 'facial_features: incomplete horizontal movement of the eyes to the bilateral side'], 'MSK': ['muscle: his upper and lower limbs showed no paralysis, they exhibited severe cerebellar ataxia and hypotonia.'], 'ENDO': ['endocrine_glands: Although the thyrotropin-releasing drug taltirelin did not relieve his symptoms'], 'LYMPH': [], 'History': ['past_medical_history: optic neuropathy (mother and uncle)', 'past_medical_history: subarachnoid hemorrhage (mother)', 'history_of_present_illness: visual and gait disturbances first appeared at 10 years of age', 'history_of_present_illness: horizontal gaze palsy, gaze-evoked nystagmus, dysarthria, cerebellar ataxia', 'history_of_present_illness: mild disturbance of cognitive function (Revised Wechsler Adult Intelligence Scale: total IQ=73, performance IQ=58, verbal IQ=91)', 'history_of_present_illness: bilateral exotropia, double vision, incomplete horizontal movement of the eyes to the bilateral side, horizontal and vertical gaze-evoked nystagmus', 'history_of_present_illness: severe cerebellar ataxia and hypotonia in upper and lower limbs', 'family_and_genetics_history: optic neuropathy (mother and uncle)', 'family_and_genetics_history: subarachnoid hemorrhage (mother)', 'chief_complaint: severe dizziness and double vision', 'family_and_genetics_history: The G11778A mutation, which causes an Arg-to-His amino acid substitution in ND4, was also observed.', 'family_and_genetics_history: PCR-RFLP data showed that this mutation in the patient and his mother was heteroplasmic.', 'family_and_genetics_history: The semiquantitative analysis performed to determine the effect of this mutation on disease severity revealed that III-1 had a 92% heteroplasmic G11778A mutation, and II-2 had a 70% heteroplasmic G11778A mutation.',

	'family_and_genetics_history: The mutations in mtDNA include homoplasmic T3 394C mutation of the mtDNA which causes a Tyr-to-His amino acid substitution i n ND1.'], 'Lab_Image': ['Lab_Image: G11778A mutation heteroplasmic levels: III-1 (92%), I I-2 (70%)', 'Lab_Image: homoplasmic T3394C mutation of the mtDNA', 'Lab_Image: The G11778A mutation, which causes an Arg-to-His amino acid subs titution in ND4, was also observed.', 'Lab_Image: PCR-RFLP data showed that this mutation in the patient and his mot her was heteroplasmic.', 'Lab_Image: Semiquantitative analysis performed to determine the effect of this mutation on disease severity revealed that III-1 had a 92% heteroplasmic G11778 A mutation, and II-2 had a 70% heteroplasmic G11778A mutation.', 'Lab_Image: The T3394C mutation causes a Tyr-to-His amino acid substitution in ND1.', 'Lab_Image: PCR-RFLP data revealed that the T3394C mutation present in both t he proband (III-1) and his mother (II-2) was homoplasmic.', 'Lab_Image: Brain and orbit MRI disclosed atrophy of the optic nerve and cerebel lum', 'Lab_Image: Orbital MRI revealed atrophy of the optic nerve', 'Lab_Image: brain MRI disclosed severe atrophy of the cerebellum and mild atro phy of the brain stem', 'Lab_Image: The bilateral inferior olivary nucleus exhibited low signal intensities on T1-weighted imaging, and high signal intensities on T2-weighted imaging, sug gesting degeneration', 'Lab_Image: PCR products that included the previously reported candidate abnor mal points were analyzed by capillary electrophoresis using an automated DNA s equencer.', 'Lab_Image: The G11778A and T3394C mutations were identified, while the A32 43G mutation was not detected.', 'Lab_Image: Sequence analysis revealed the homoplasmic T3394C mutation of th e mtDNA', 'Lab_Image: PCR-RFLP data revealed that the T3394C mutation present in both t he proband (III-1) and his mother (II-2) was homoplasmic; differences between t he patient and his mother were not observed for this mutation.', 'Lab_Image: The G11778A mutation, which causes an Arg-to-His amino acid subs titution in ND4, was also observed', 'Lab_Image: PCR-RFLP data showed that this mutation in the patient and his mot her was heteroplasmic.', 'Lab_Image: PCR products that included the previously reported candidate abnor mal points were analyzed by capillary electrophoresis using an automated DNA s equencer.', 'Lab_Image: The G11778A and T3394C mutations were identified, while the A32 43G mutation was not detected.', 'Lab_Image: PCR-RFLP data revealed that the T3394C mutation present in both t he proband (III-1) and his mother (II-2) was homoplasmic; differences between t he patient and his mother were not observed for this mutation.', 'Lab_Image: The G11778A mutation, which causes an Arg-to-His amino acid subs titution in ND4, was also observed.', 'Lab_Image: PCR-RFLP data showed that this mutation in the patient and his mot her was heteroplasmic.', 'Lab_Image: Ophthalmological examination revealed atrophy of the optic nerve, but there were no pigmentation changes of the retina.', 'Lab_Image: Orbital MRI revealed atrophy of the optic nerve.', 'Lab_Image: MRI of his mother disclosed mild atrophy of the optic nerve.']}

GPT-4o Few-Shot Prompts Category-Based Filtering and Targeted Extraction	{'Neuro': ['symptoms: horizontal gaze palsy', 'symptoms: gaze-evoked nystagmus', 'symptoms: dysarthria', 'symptoms: cerebellar ataxia', 'symptoms: bilateral exotropia', 'symptoms: double vision', 'symptoms: incomplete horizontal eye movement', 'symptoms: tremor in neck', 'symptoms: severe cerebellar ataxia', 'symptoms: hypotonia', 'mild disturbance: True', 'total IQ: 73', 'performance IQ: 58', 'verbal IQ: 91', 'I: no disturbances', 'VII: no disturbances', 'VIII: no disturbances', 'XII: no disturbances', 'tremor: neck', 'palatal myoclonus: not observed', 'deep_tendon_reflex: normal', 'sensory_system: normal', 'optic_nerve: atrophy', 'cerebellum: severe atrophy', 'brain_stem: mild atrophy', 'T1-weighted: low signal intensities', 'T2-weighted: high signal intensities', 'diagnosis: LHON plus olivocerebellar degeneration', 'mitochondrial_DNA_mutations: heteroplasmic G11778A', 'mitochondrial_DNA_mutations: homoplasmic T3394C', 'optic_neuropathy: True', 'subarachnoid_hemorrhage: True', 'optic_nerve: mild atrophy', 'pons: mild atrophy', 'cerebellum: mild atrophy', 'inferior_olivary_nucleus: no signal changes', 'optic_neuropathy: True', 'other_neurological_abnormalities: not observed', 'detailed_clinical_information: unavailable'], 'RESP': ['observations: No specific respiratory system observations or abnormalit ies were reported in the text.'], 'GU': ['patient_age: 37', 'patient_gender: male', 'diagnostic_data: No specific genitourinary system diagnostic data provided in th e text.', 'optic_nerve: atrophy', 'cerebellum: severe atrophy', 'brain_stem: mild atrophy', 'inferior_olivary_nucleus: degenerative changes with low signal intensities on T1 -weighted imaging and high signal intensities on T2-weighted imaging', 'mitochondrial_DNA_mutations: heteroplasmic G11778A', 'mitochondrial_DNA_mutations: homoplasmic T3394C', 'optic_neuropathy: True', 'subarachnoid_hemorrhage: True',

	'optic_nerve: mild atrophy', 'pons: mild atrophy', 'cerebellum: mild atrophy', 'inferior_olivary_nucleus: no signal changes', 'optic_neuropathy: True', 'other_neurological_abnormalities: False'], 'DERM': ['patient_age: 37', 'gender: male', 'diagnosis: LHON plus olivocerebellar degeneration', 'symptoms: visual disturbances', 'symptoms: gait disturbances', 'symptoms: horizontal gaze palsy', 'symptoms: gaze-evoked nystagmus', 'symptoms: dysarthria', 'symptoms: cerebellar ataxia', 'symptoms: severe dizziness', 'symptoms: double vision', 'symptoms: bilateral exotropia', 'symptoms: incomplete horizontal eye movement', 'symptoms: tremor in neck', 'symptoms: hypotonia', 'total_IQ: 73', 'performance_IQ: 58', 'verbal_IQ: 91', 'I: normal', 'VII: normal', 'VIII: normal', 'XII: normal', 'deep_tendon_reflex: normal', 'sensory_system: normal', 'optic_nerve: atrophy', 'retina: no pigmentation changes', 'orbital_MRI: atrophy of the optic nerve', 'cerebellum: severe atrophy', 'brain_stem: mild atrophy', 'T1_weighted: low signal intensities', 'T2_weighted: high signal intensities', 'mitochondrial_DNA_mutations: heteroplasmic G11778A', 'mitochondrial_DNA_mutations: homoplasmic T3394C', 'optic_neuropathy: True', 'subarachnoid_hemorrhage: True', 'optic_nerve: mild atrophy', 'pons: mild atrophy', 'cerebellum: mild atrophy', 'inferior_olivary_nucleus: no signal changes', 'optic_neuropathy: True', 'other_neurological_abnormalities: False'], 'MSK': ['observations: Severe cerebellar ataxia', 'observations: Hypotonia in upper and lower limbs'], 'ENDO': ['age: 37', 'gender: male', 'symptoms: visual disturbances', 'symptoms: gait disturbances', 'symptoms: horizontal gaze palsy', 'symptoms: gaze-evoked nystagmus',

	'symptoms: dysarthria', 'symptoms: cerebellar ataxia', 'symptoms: severe dizziness', 'symptoms: double vision', 'symptoms: bilateral exotropia', 'symptoms: incomplete horizontal eye movement', 'symptoms: tremor in neck', 'symptoms: hypotonia', 'total_IQ: 73', 'performance_IQ: 58', 'verbal_IQ: 91', 'cranial_nerves: No disturbances in cranial nerves I, VII, VIII, and XII', 'deep_tendon_reflex: No abnormal findings', 'sensory_system: No abnormal findings', 'optic_nerve: atrophy', 'retina: no pigmentation changes', 'orbital_MRI: atrophy of the optic nerve', 'cerebellum: severe atrophy', 'brain_stem: mild atrophy', 'T1_weighted: low signal intensities', 'T2_weighted: high signal intensities', 'diagnosis: LHON plus olivocerebellar degeneration', 'mitochondrial_DNA: heteroplasmic G11778A mutation', 'mitochondrial_DNA: homoplasmic T3394C mutation', 'conditions: optic neuropathy', 'conditions: subarachnoid hemorrhage', 'optic_nerve: mild atrophy', 'pons: mild atrophy', 'cerebellum: mild atrophy', 'inferior_olivary_nucleus: No signal changes', 'conditions: optic neuropathy'], 'LYMPH': ['age: 37', 'gender: male', 'symptoms: visual disturbances', 'symptoms: gait disturbances', 'symptoms: horizontal gaze palsy', 'symptoms: gaze-evoked nystagmus', 'symptoms: dysarthria', 'symptoms: cerebellar ataxia', 'symptoms: severe dizziness', 'symptoms: double vision', 'symptoms: bilateral exotropia', 'symptoms: incomplete horizontal eye movement', 'symptoms: tremor in neck', 'symptoms: hypotonia', 'total_IQ: 73', 'performance_IQ: 58', 'verbal_IQ: 91', 'cranial_nerves: No disturbances in cranial nerves I, VII, VIII, and XII', 'deep_tendon_reflex: No abnormal findings', 'sensory_system: No abnormal findings', 'optic_nerve: atrophy', 'retina: no pigmentation changes', 'orbital_MRI: atrophy of the optic nerve', 'cerebellum: severe atrophy',

	'brain_stem: mild atrophy', 'T1_weighted: low signal intensities', 'T2_weighted: high signal intensities', 'diagnosis: LHON plus olivocerebellar degeneration', 'mitochondrial_DNA: heteroplasmic G11778A mutation', 'mitochondrial_DNA: homoplasmic T3394C mutation', 'conditions: optic neuropathy', 'conditions: subarachnoid hemorrhage', 'optic_nerve: mild atrophy', 'pons: mild atrophy', 'cerebellum: mild atrophy', 'inferior_olivary_nucleus: no signal changes', 'conditions: optic neuropathy', 'clinical_information: unable to confirm'], 'History': ['chief_complaint: Severe dizziness and double vision', 'age_of_onset: 10', 'initial_symptoms: visual disturbances', 'initial_symptoms: gait disturbances', 'current_symptoms: horizontal gaze palsy', 'current_symptoms: gaze-evoked nystagmus', 'current_symptoms: dysarthria', 'current_symptoms: cerebellar ataxia', 'current_symptoms: severe dizziness', 'current_symptoms: double vision', 'neurological_findings: mild cognitive function disturbance', 'neurological_findings: bilateral exotropia', 'neurological_findings: double vision', 'neurological_findings: incomplete horizontal eye movement', 'neurological_findings: horizontal and vertical gaze-evoked nystagmus', 'neurological_findings: dysarthria', 'neurological_findings: neck tremor', 'neurological_findings: severe cerebellar ataxia', 'neurological_findings: hypotonia', 'optic_nerve_atrophy: True', 'cerebellum_atrophy: True', 'brain_stem_atrophy: mild', 'inferior_olivary_nucleus_degeneration: True', 'diagnosis: LHON plus olivocerebellar degeneration', 'mutational_analyses: heteroplasmic G11778A', 'mutational_analyses: homoplasmic T3394C mutations', 'conditions: optic neuropathy', 'conditions: subarachnoid hemorrhage', 'optic_nerve_atrophy: mild', 'pons_atrophy: mild', 'cerebellum_atrophy: mild', 'conditions: optic neuropathy', 'clinical_information: not confirmed'], 'Lab_Image': []}
GPT-4o Few-shot prompts	{'iem': ['iem: is_inborn_error_of_metabolism', 'iem: reasoning'], 'Pregnancy': ['mutation_type: homoplasmic', 'amino_acid_change: Tyr-to-His', 'gene: ND1', 'mutation_type: heteroplasmic', 'amino_acid_change: Arg-to-His',

Uniform Application of Category Prompts	'gene: ND4', 'heteroplasmy_percentage: 70'], 'Neuro': ['symptoms: horizontal gaze palsy', 'symptoms: gaze-evoked nystagmus', 'symptoms: dysarthria', 'symptoms: cerebellar ataxia', 'symptoms: bilateral exotropia', 'symptoms: double vision', 'symptoms: incomplete horizontal eye movement', 'symptoms: tremor in neck', 'symptoms: severe cerebellar ataxia', 'symptoms: hypotonia', 'mild disturbance: True', 'total IQ: 73', 'performance IQ: 58', 'verbal IQ: 91', 'I: no disturbances', 'VII: no disturbances', 'VIII: no disturbances', 'XII: no disturbances', 'tremor: neck', 'palatal myoclonus: not observed', 'deep_tendon_reflex: normal', 'sensory_system: normal', 'optic_nerve: atrophy', 'cerebellum: severe atrophy', 'brain_stem: mild atrophy', 'T1-weighted: low signal intensities', 'T2-weighted: high signal intensities', 'diagnosis: LHON plus olivocerebellar degeneration', 'mitochondrial_DNA_mutations: heteroplasmic G11778A', 'mitochondrial_DNA_mutations: homoplasmic T3394C', 'optic_neuropathy: True', 'subarachnoid_hemorrhage: True', 'optic_nerve: mild atrophy', 'pons: mild atrophy', 'cerebellum: mild atrophy', 'inferior_olivary_nucleus: no signal changes', 'optic_neuropathy: True', 'other_neurological_abnormalities: not observed', 'detailed_clinical_information: unavailable', 'Neuro: {\n "neurological": {\n "genetic_mutations": {\n "G11778A": {\n "gene": "ND4",\n "mutation_type": "heteroplasmic",\n "amino_acid_change": "Arg-to-His",\n "patient_percentage": 92,\n "mother_percentage": 70\n },\n "T3394C": {\n "gene": "ND1",\n "mutation_type": "homoplasmic",\n "amino_acid_change": "Tyr-to-His"\n }\n },\n "associated_conditions": [\n "Leber\'s Hereditary Optic Neuropathy (LHON)",\n "mitochondrial encephalomyopathy, lactic acidosis, and stroke-like episodes"\n],\n "excluded_conditions": [\n "cerebellar ataxia",\n "polyglutamine diseases"\n]\n },\n "cognitive": {},\n "head": {}\n}'], 'CVS': ['CVS: {\n "cardiovascular_system": {\n "heart_function": "No specific information provided",\n "blood_vessel_function": "No specific information provided"\n },\n "vascular_system": {\n "blood_vessel_conditions": "The patient\'s mother has a history of subarachnoid hemorrhage."\n }\n}'],

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	'symptoms: cerebellar ataxia', 'symptoms: severe dizziness', 'symptoms: double vision', 'symptoms: bilateral exotropia', 'symptoms: incomplete horizontal eye movement', 'symptoms: tremor in neck', 'symptoms: hypotonia', 'total_IQ: 73', 'performance_IQ: 58', 'verbal_IQ: 91', 'I: normal', 'VII: normal', 'VIII: normal', 'XII: normal', 'deep_tendon_reflex: normal', 'sensory_system: normal', 'optic_nerve: atrophy', 'retina: no pigmentation changes', 'orbital_MRI: atrophy of the optic nerve', 'cerebellum: severe atrophy', 'brain_stem: mild atrophy', 'T1_weighted: low signal intensities', 'T2_weighted: high signal intensities', 'mitochondrial_DNA_mutations: heteroplasmic G11778A', 'mitochondrial_DNA_mutations: homoplasmic T3394C', 'optic_neuropathy: True', 'subarachnoid_hemorrhage: True', 'optic_nerve: mild atrophy', 'pons: mild atrophy', 'cerebellum: mild atrophy', 'inferior_olivary_nucleus: no signal changes', 'optic_neuropathy: True', 'other_neurological_abnormalities: False'], 'MSK': ['observations: Severe cerebellar ataxia', 'observations: Hypotonia in upper and lower limbs'], 'ENDO': ['age: 37', 'gender: male', 'symptoms: visual disturbances', 'symptoms: gait disturbances', 'symptoms: horizontal gaze palsy', 'symptoms: gaze-evoked nystagmus', 'symptoms: dysarthria', 'symptoms: cerebellar ataxia', 'symptoms: severe dizziness', 'symptoms: double vision', 'symptoms: bilateral exotropia', 'symptoms: incomplete horizontal eye movement', 'symptoms: tremor in neck', 'symptoms: hypotonia', 'total_IQ: 73', 'performance_IQ: 58', 'verbal_IQ: 91', 'cranial_nerves: No disturbances in cranial nerves I, VII, VIII, and XII', 'deep_tendon_reflex: No abnormal findings', 'sensory_system: No abnormal findings',

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GPT-4o Few-Shot Prompt System-Wide Unified Extraction	{'patient': ['patient: age', 'patient: gender', 'patient: symptoms', 'patient: neurological_examination', 'patient: ophthalmological_examination', 'patient: imaging', 'patient: diagnosis', 'patient: genetic_analysis'], 'family_history': ['family_history: mother', 'family_history: uncle', 'family_history: grandmother'], 'Lab_Image': []}
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GPT-4o Zero-shot prompts Uniform Application of Category Prompts	{'iem': ['iem: is_inborn_error_of_metabolism', 'iem: reasoning'], 'Pregnancy': ['mutation_type: homoplasmic', 'amino_acid_change: Tyr-to-His', 'gene: ND1', 'mutation_type: heteroplasmic', 'amino_acid_change: Arg-to-His', 'gene: ND4', 'heteroplasmy_percentage: 70'], 'Neuro': ['symptoms: horizontal gaze palsy', 'symptoms: gaze-evoked nystagmus', 'symptoms: dysarthria', 'symptoms: cerebellar ataxia', 'symptoms: bilateral exotropia', 'symptoms: double vision', 'symptoms: incomplete horizontal eye movement', 'symptoms: tremor in neck', 'symptoms: severe cerebellar ataxia', 'symptoms: hypotonia', 'mild disturbance: True', 'total IQ: 73', 'performance IQ: 58', 'verbal IQ: 91',

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	'symptoms: bilateral exotropia', 'symptoms: incomplete horizontal eye movement', 'symptoms: tremor in neck', 'symptoms: hypotonia', 'total_IQ: 73', 'performance_IQ: 58', 'verbal_IQ: 91', 'cranial_nerves: No disturbances in cranial nerves I, VII, VIII, and XII', 'deep_tendon_reflex: No abnormal findings', 'sensory_system: No abnormal findings', 'optic_nerve: atrophy', 'retina: no pigmentation changes', 'orbital_MRI: atrophy of the optic nerve', 'cerebellum: severe atrophy', 'brain_stem: mild atrophy', 'T1_weighted: low signal intensities', 'T2_weighted: high signal intensities', 'diagnosis: LHON plus olivocerebellar degeneration', 'mitochondrial_DNA: heteroplasmic G11778A mutation', 'mitochondrial_DNA: homoplasmic T3394C mutation', 'conditions: optic neuropathy', 'conditions: subarachnoid hemorrhage', 'optic_nerve: mild atrophy', 'pons: mild atrophy', 'cerebellum: mild atrophy', 'inferior_olivary_nucleus: No signal changes', 'conditions: optic neuropathy', 'mutation: G11778A', 'gene: ND4', 'amino_acid_change: Arg-to-His', 'heteroplasmic: True', 'patient_heteroplasmy_percentage: 92', 'mother_heteroplasmy_percentage: 70', 'mutation: T3394C', 'gene: ND1', 'amino_acid_change: Tyr-to-His', 'homoplasmic: True', 'mutations_not_detected: A3243G', 'cerebellar_ataxia_associated_abnormalities: Not found'], 'LYMPH': ['age: 37', 'gender: male', 'symptoms: visual disturbances', 'symptoms: gait disturbances', 'symptoms: horizontal gaze palsy', 'symptoms: gaze-evoked nystagmus', 'symptoms: dysarthria', 'symptoms: cerebellar ataxia', 'symptoms: severe dizziness', 'symptoms: double vision', 'symptoms: bilateral exotropia', 'symptoms: incomplete horizontal eye movement', 'symptoms: tremor in neck', 'symptoms: hypotonia', 'total_IQ: 73', 'performance_IQ: 58',

	'verbal_IQ: 91', 'cranial_nerves: No disturbances in cranial nerves I, VII, VIII, and XII', 'deep_tendon_reflex: No abnormal findings', 'sensory_system: No abnormal findings', 'optic_nerve: atrophy', 'retina: no pigmentation changes', 'orbital_MRI: atrophy of the optic nerve', 'cerebellum: severe atrophy', 'brain_stem: mild atrophy', 'T1_weighted: low signal intensities', 'T2_weighted: high signal intensities', 'diagnosis: LHON plus olivocerebellar degeneration', 'mitochondrial_DNA: heteroplasmic G11778A mutation', 'mitochondrial_DNA: homoplasmic T3394C mutation', 'conditions: optic neuropathy', 'conditions: subarachnoid hemorrhage', 'optic_nerve: mild atrophy', 'pons: mild atrophy', 'cerebellum: mild atrophy', 'inferior_olivary_nucleus: no signal changes', 'conditions: optic neuropathy', 'clinical_information: unable to confirm', 'gene: ND4', 'mutation_type: heteroplasmic', 'amino_acid_change: Arg-to-His', 'patient_heteroplasmy_percentage: 92', 'mother_heteroplasmy_percentage: 70', 'gene: ND1', 'mutation_type: homoplasmic', 'amino_acid_change: Tyr-to-His', 'patient_status: present', 'mother_status: present', 'gene: ND1', 'mutation_type: not detected', 'cerebellar_ataxia: not found', 'polyglutamine_diseases: not found'], 'History': ['chief_complaint: Severe dizziness and double vision', 'age_of_onset: 10', 'symptoms_at_onset: visual disturbances', 'symptoms_at_onset: gait disturbances', 'current_symptoms: horizontal gaze palsy', 'current_symptoms: gaze-evoked nystagmus', 'current_symptoms: dysarthria', 'current_symptoms: cerebellar ataxia', 'current_symptoms: severe dizziness', 'current_symptoms: double vision', 'cognitive_function: mild disturbance', 'total_IQ: 73', 'performance_IQ: 58', 'verbal_IQ: 91', 'neurological_disturbances: bilateral exotropia', 'neurological_disturbances: double vision', 'neurological_disturbances: incomplete horizontal eye movement', 'neurological_disturbances: horizontal and vertical gaze-evoked nystagmus', 'neurological_disturbances: dysarthria',

	'other_findings: tremor in neck', 'other_findings: severe cerebellar ataxia', 'other_findings: hypotonia', 'optic_nerve: atrophy', 'retina: no pigmentation changes', 'orbital_MRI: atrophy of the optic nerve', 'cerebellum: severe atrophy', 'brain_stem: mild atrophy', 'inferior_olivary_nucleus: degeneration', 'mitochondrial_DNA_mutations: heteroplasmic G11778A', 'mitochondrial_DNA_mutations: homoplasmic T3394C', 'conditions: optic neuropathy', 'conditions: subarachnoid hemorrhage', 'optic_nerve: mild atrophy', 'pons: mild atrophy', 'cerebellum: mild atrophy', 'conditions: optic neuropathy', 'clinical_information: not confirmed', 'proband: III-1', 'mother: II-2', 'mutation: G11778A', 'gene: ND4', 'amino_acid_substitution: Arg-to-His', 'proband: 92', 'mother: 70', 'mutation: T3394C', 'gene: ND1', 'amino_acid_substitution: Tyr-to-His', 'homoplasmic: True', 'other_genetic_conditions: No established genetic abnormalities associated with cerebellar ataxia including polyglutamine diseases were found.'], 'Lab_Image': []}
GPT-4o Zero-shot prompts System-Wide Unified Extraction	{'Patient': ['Patient: Age', 'Patient: Gender', 'Patient: Symptoms', 'Patient: Neurological Examination', 'Patient: Ophthalmological Examination', 'Patient: Imaging', 'Patient: Blood and Cerebrospinal Fluid Analyses', 'Patient: Exercise Test', 'Patient: Diagnosis', 'Patient: Genetic Findings'], 'Family History': ['Family History: Mother (II-2)', 'Family History: Uncle (II-3)', 'Family History: Grandmother (I-2)'], 'Lab_Image': []}