

RARE / INHERITED DISEASE FRAMING

Recurrent swelling and abdominal attacks: phenotype-led research

Illustrative fictional composite. This sample demonstrates report structure and reasoning style; it is not a real patient outcome.

CLINICIAN QUICK REVIEW

The history contains a high-value pattern that routine 'allergy' framing does not fully explain: recurrent abdominal crises, swelling episodes without a consistent itchy rash, and a similar pattern in a first-degree relative. This does not establish hereditary angioedema, but it is strong enough to justify a focused branch rather than another broad rare-disease list.

Research objective

Does the pattern justify moving beyond common allergic explanations toward a bradykinin-mediated or inherited angioedema pathway, and what evidence would most efficiently discriminate that branch?

Outcome strategy tree

Branch	Patient-specific rationale	What conflicts / is missing	Best discriminating next evidence
Bradykinin-mediated / inherited angioedema family	Episodic swelling + abdominal attacks + family pattern; standard allergy treatment reportedly unreliable.	No contemporaneous examination during an attack; complement/C1-inhibitor status not available in this fictional record.	Retrieve or obtain targeted complement/C1-inhibitor evidence and document attack phenotype/timing.
Histamine-mediated angioedema / allergy	Swelling is a common phenotype and some attacks followed nonspecific exposures.	Repeated abdominal crises and absent urticaria in several attacks are less typical; family clustering needs explanation.	Clarify urticaria/pruritus, response to antihistamine/epinephrine, and exposure reproducibility.
Grouped mimics	Medication-related, acquired complement disorders, mast-cell conditions and other mimics remain possible depending on age/exposures.	Current fictional history is insufficient to rank them individually.	Use age of onset, medication chronology, associated findings and focused labs to decide whether any mimic deserves its own branch.

Actionable next moves

- Do not order a broad genetic panel first. Confirm that the phenotype and first-line biochemical pathway justify genetic escalation.
- Ask the patient to bring photographs/records from an attack if available and reconstruct response to prior emergency treatments.
- If targeted evidence supports an inherited pathway, use the phenotype and family history to guide confirmatory specialist/genetic evaluation.

Question-and-answer transcript summary

Q: During attacks, is there usually an itchy rash or hives?

A: The patient reports swelling and abdominal pain, but several major attacks occurred without an itchy rash.

Q: Did standard allergy treatment reliably stop attacks?

A: The patient recalls inconsistent or minimal benefit; documentation is incomplete.

Q: Has anyone in the family had similar unexplained swelling or abdominal crises?

A: A parent reportedly had recurrent swelling episodes but was never formally diagnosed.

Detailed case synthesis

The central value of the extended interview is pattern reconstruction. Individual symptoms are common; the combination, recurrence, treatment response and family pattern changes the information value. The research fellow therefore narrows the next step to a small number of discriminating observations rather than presenting a long differential.

Rare, inherited and multisystem considerations

The inherited-angioedema family is the main rare-disease branch because the fictional phenotype provides a specific reason to consider it. Other rare causes should remain grouped unless new evidence gives them patient-specific support.

Therapeutic or registered-trial opportunities

No therapeutic trial or registered trial is proposed from this fictional evidence. If a specific molecular diagnosis were later established and standard care were inadequate, current registered studies could be searched for clinician review; no eligibility would be inferred.

What could change our mind?

A new examination finding, a prior result that changes chronology, a clearly documented treatment response, or a targeted test that directly discriminates between the leading branches should trigger revision rather than simply adding more diagnoses.

Reopen plan

Reopen the same case when the highest-value requested evidence arrives or when treatment response materially changes the working model.

References

In a live case, WhatDx would cite the patient records used and add authoritative literature or official trial-registry sources where current external facts materially affect the strategy.