

## A Girl with Severe Hand Swelling and Abdominal Cramps

Arturo Borzutzky,<sup>1</sup> Lynda C. Schneider,<sup>1</sup> and Raif S. Geha<sup>1</sup>

<sup>1</sup>Division of Immunology, Children's Hospital Boston, Harvard Medical School, Boston, MA.

Address correspondence to this author at: Children's Hospital Boston, Division of Immunology, 300 Longwood Avenue, Boston, MA 02115. Fax 617-730-0249; e-mail arturo.borzutzky@childrens.harvard.edu.

### CASE DESCRIPTION

A 16-year-old girl presented to the emergency room with severe swelling of her left hand and forearm. Her symptoms began 12 h earlier with left forearm swelling that progressed to the tips of her fingers. She reported her forearm felt normal, but her hand had a tingling sensation and she was barely able to move her fingers. She also reported having had moderately painful abdominal cramps earlier that day. She denied urticarial or pruritic rashes, respiratory distress, and throat tightness. During the previous week she had felt stressed because of schoolwork and also had developed a mild headache, clear rhinorrhea, and cough. She had never had episodes of swelling in the past.

Family history revealed similar episodes in multiple family members. The patient's father and 15-year-old sister had histories of numerous episodes of painful localized edema since childhood. Her paternal grandmother, great-grandfather, and great-great-grandfather died of asphyxiation due to attacks of angioedema and swelling of the throat. Her mother and 13-year-old brother were healthy.

On examination, the patient appeared well and in no respiratory distress. Vital signs were stable. Her skin had no rashes. Her left upper extremity was severely swollen from the midforearm to the fingertips. Her left hand was cold and capillary refill was mildly delayed. The patient reported a tingling sensation when her fingers were superficially palpated. Flexion and extension of her fingers were greatly reduced. The patient's abdomen was soft but diffusely tender, without hepatosplenomegaly. There was no swelling of other body parts. Physical examination results were otherwise unremarkable.

Complete blood count showed mild leukocytosis with neutrophilic predominance. C-reactive protein was 15 mg/L (reference interval <5 mg/L), C3 was 1170 mg/L (reference interval 830–1770 mg/L), and C4 was <60 mg/L (reference interval 140–420 mg/L).

Questions to Consider
• What is the most likely diagnosis in our patient?
• What laboratory tests would you order to confirm the diagnosis?
• Given the family history, what inheritance pattern would you assume is most likely to explain our patient's condition?
• Why does our patient have a low C4?

## Final Publication and Comments

The final published version with discussion and comments from the experts will appear in the October 2008 issue of *Clinical Chemistry*. To view the case and comments online, go to <http://www.clinchem.org/content/vol54/issue10/> and follow the link to the Clinical Case Study and Commentaries.

## Educational Centers

If you are associated with an educational center and would like to receive the cases and questions 2-3 weeks in advance of publication, please email [clinchem@aacc.org](mailto:clinchem@aacc.org).

AACC is pleased to allow free reproduction and distribution of this Clinical Case Study for personal or classroom discussion use. When photocopying, please make sure the DOI and copyright notice appear on each copy.

All previous Clinical Cases Studies can be accessed and downloaded online at <http://www.aacc.org/resourcecenters/casestudies/>.

---

AACC is a leading professional society dedicated to improving healthcare through laboratory medicine. Its nearly 10,000 members are clinical laboratory professionals, physicians, research scientists, and others involved in developing tests and directing laboratory operations. AACC brings this community together with programs that advance knowledge, expertise, and innovation. AACC is best known for the respected scientific journal, *Clinical Chemistry*, the award-winning patient-centered web site *Lab Tests Online*, and the world's largest conference on laboratory medicine and technology. Through these and other programs, AACC advances laboratory medicine and the quality of patient care.