

# Understanding the Rare Disorder Community

## Top Facts ER Staff Should Know About People with Rare Disorders

Adapted from E. Migdol, "13 Things ER Staff Should Know About People with Rare Diseases" *The Mighty*

1. Accept that rare disorder patients and/or their parents know more about their illness than you do.
2. Call the specialists if you need help.
3. Believe parents when they explain their child's condition and medical history.
4. Ultimately, listening to the person with a rare disease and having a compassionate, patient attitude goes a long way, even if you are unfamiliar with the specific condition.



## Emergency Tips when Caring for an Individual with Cornelia de Lange Syndrome (CdLS)

Provided by, Antonie Kline, M.D., Medical Director for the CdLS Foundation

- With sudden and acute abdominal pain, rigid abdomen, consider **volvulus** and/or **bowel obstruction** at any age
- When presenting with respiratory symptoms and/or pneumonia, consider **gastroesophageal reflux** and aspiration as a cause
- Seizures can occur at any age
- Pyloric stenosis has an increased incidence
- Sudden change in behavior with increased self-injury and aggression may indicate an underlying organic cause
- Anesthesia can be difficult due to small mouth and chin. We **do not** recommend using Versed for sedation due to reactions.

Visit our website to learn more about Critical Care  
[www.CdLSusa.org/critical-care](http://www.CdLSusa.org/critical-care)

