Familial Dysautonomia

What is FD?

- Familial Dysautonomia (FD), also known as Riley-Day Syndrome, is a rare genetic disorder that affects sensory and autonomic nerves
- FD is a recessive inherited disorder – both parents must have the mutated gene for a child to be born with FD. People may be carriers but have no symptoms.
- Mutations in this gene cause both sensory and autonomic nerves to not develop properly, leading to a decrease in sensitivity to pain, as well as severely abnormal autonomic nervous system functions
- Symptoms are usually present at birth and, while survival rates have improved, it is still one of the few forms of dysautonomia that are fatal. Symptoms and severity vary between patients, but the condition is progressive
- Carrier frequency is much higher in Ashkenazi Jews and those of Eastern European decent, with as estimates as high as 1 in 27 being carriers

What are the symptoms?

- Patients at birth suffer from poor sucking ability, loss of muscle tone, low body temperature, and difficulty swallowing. They may also have profuse sweating and crying without tears
- Episodes of autonomic crisis include cyclical vomiting, severe hypertension, fast heart rates, and elevated body temperature
- Other symptoms include: poor/slow growth and development, drooling, decreased or no sensation of pain or temperature, excessive sweating, blotchy-reddening of skin during feeding or excitement, lack of taste buds, spinal curvature that leads to restrictive lung disease and frequent lung infections, GERD, sleep apnea, abnormal renal function, fainting, and abnormal heart rhythms

For more information visit:
Familial Dysautonomia Foundation at http://www.familialdysautonomia.org/
FD Now at https://fdnow.org/

Additional sources:
NORD: https://rarediseases.org/rare-diseases/dysautonomia-familial

NYU Langone Health: https://nyulangone.org/conditions/familial-dysautonomia

How is it diagnosed?

- Diagnosis is made through genetic testing – looking for 2 mutated copies of the IKBKAP gene
- Prenatal testing in high-risk populations may be beneficial to determine if parents are carriers

How is it treated?

- There is no cure for FD, only symptom management, supportive therapies, and preventative measures for complications
- Management is individually focused to address blood pressure and heart rate management, breathing problems, feeding and digestive difficulties, and kidney function
- Patients may also need speech and/or occupational therapy to address development or growth delays and to prevent injury from joint, muscle, or skeletal complications. Patients may also need assistive devices like wheelchairs, walkers, canes, or braces.