

# Pregnancy Complicated by Familial Dysautonomia Riley-Day Syndrome: A Case Report

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## Abstract

To illustrate the importance of developing a multidisciplinary approach when managing a pregnant patient diagnosed with Familial Dysautonomia Riley-Day syndrome. A multisystem dysfunction results from this disorder stemming from the progressive nature of the disease affecting both the sensory and autonomic nervous system. A 27 year old female with known Familial Dysautonomia Riley-Day syndrome presented to High Risk Obstetrical Clinic for prenatal care with progressive sensory and autonomic dysfunction since her previous delivery five years ago. The patient delivered a healthy infant at 36 weeks gestation requiring no intervention by the Neonatal Intensive Care Unit. Both mother and infant were discharged home together on post-operative day 4. Through the utilization of a multidisciplinary approach both prenatally and post-partum, a pregnant patient with Familial Dysautonomia Riley-Day syndrome experiencing multiple serious medical problems can successfully deliver an infant requiring no additional medical support. Ideally both mother and infant can be discharged home together.

**Keywords:** Familial dysautonomia Riley-Day syndrome; Sensory and autonomic dysfunction; Pregnancy; Multidisciplinary approach

## Introduction

Familial dysautonomia Riley-Day syndrome is one of several hereditary types of neurologic disorders classified under hereditary sensory autonomic neuropathies (HSAN) [1-8]. This classification manifests itself in varying degrees of sensory and autonomic dysfunction [1-8]. Familial dysautono-

mia Riley-Day syndrome or HSAN III is the most common of these disorders affecting individuals almost exclusively of Ashkenazi Jewish ancestry [1-8]. This case involves a patient diagnosed with the neuropathy that successfully delivered a healthy infant using a multidisciplinary approach. Despite the severity of her medical condition pre and post delivery, both she and her infant were discharged requiring no additional hospital days.

## Case Report

A 27 year old white female of Ashkenazi descent G2P1 was referred to the High Risk Clinic with an established diagnosis of familial dysautonomia Riley-Day syndrome. Five years prior, she was delivered by cesarean section at another institution at 31 weeks. Her prenatal course then was complicated by preterm labor, gestational diabetes and pneumonia. The indication for the cesarean performed under epidural anesthesia was fetal distress. An infant weighing 3#14 ounces was delivered who required several weeks in the Neonatal Intensive Care prior to discharge.

Consistent with her diagnosis of Familial Dysautonomia Riley-Day Syndrome, this patient presented for prenatal care with several significant medical problems. Two years preceding the current pregnancy, she was referred to the New York University Dysautonomia Center secondary to a notable increase in the frequency and severity of her autonomic crises along with pronounced fatigue. At her initial prenatal visit, she complained of experiencing both severe orthostatic hypotension in addition to periods of marked hypertension. Her marked shifts in blood pressure were accompanied by recurrent episodes of vomiting along with profuse sweating. She noted that stress, along with any sort of infection triggered a crisis as well. Previously, she received several blood transfusions for her chronic anemia. She also noted worsening of her previously diagnosed swallowing difficulties particularly with liquids and was treated for pneumonia due to aspiration in the past. With the onset of this pregnancy, the incidences of gastro esophageal reflux occurrences have increased as well. Her skeletal deformities resulting in scoliosis required Harrington rod placement in the past. Besides recurrent uri-

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nary tract infections, she also complained of worsening of her migraine headaches.

Prior to pregnancy, her medications included: Valium (diazepam) per rectum and on occasion a Clonidine (catapres) patch applied weekly prn for hypertension. In addition, she was taking Midodrine (proamantine) for hypotension along with Flurocortisone (florinef) daily. Her gastric reflux was managed with Nexium (esomeprazole) daily. Lyrica (pregabalin) was prescribed for pain beside prenatal vitamins along with calcium supplements.

At her initial prenatal visit, a care team involving multiple disciplines was established to address her multisystem dysfunction. She first underwent genetic counseling secondary to her medications causing potential teratogenic exposure. Due to the severity of her condition, she was advised to continue her medications taken prenatally. Maternal Fetal Medicine specialists along with Obstetrical Generalists followed her pregnancy with monthly obstetrical ultrasounds for growth, in addition to weekly fetal monitoring starting at 32 weeks. She also provided multiple urine specimens ruling out infection. Gastroenterologists managed both her swallowing difficulties which significantly worsened during the pregnancy along with a notable increase in the frequency of esophageal reflux episodes. This issue became a particular problem when the patient could not complete her one hour glucose challenge testing. She was alternatively followed with Hemoglobin A1Cs and postprandial glucoses. Hematologists were consulted to coordinate blood transfusions for anemia and a Neurologist managed her incapacitated migraine headaches. Cardiologists assisted with both hypertensive and hypotensive events requiring frequent medical adjustments.

Her prenatal course was also complicated by preterm labor treated at 29 weeks with steroids Celestone (betamethosone) intramuscularly x 2 doses. In the event of a possible preterm delivery again, she was also seen by Neonatology. Finally, a referral was made to Anesthesiology for a planned repeat cesarean section and requested tubal ligation.

The patient had been scheduled for delivery at 37 weeks however she presented at 36 weeks 2 days at an outlying hospital complaining of contractions. She was given subcutaneous Terbutalin resulting in blood pressure drop to 60/40. She then rebounded with blood pressures of 180/140 to which she received Valium prior to transfer. On admission, she had notable labile blood pressures, uterine contractions occurring every 2 - 5 minutes and a cervical exam of 4 centimeters dilatation with a Category 1 fetal monitor tracing. She was 4'9" tall weighing 119 pounds. Due to her discomfort, the patient requested delivery and a repeat cesarean section with tubal ligation was performed under spinal anesthesia. The infant weighed 2255 grams or 4#15.5 ounces with Apgars of 9 at one minute and 9 at 5 minutes. The cord ph was 7.33 and the infant required no intervention by NICU regarding respiratory, intravenous or phototherapy support during his

admission.

The patient herself was transferred to the Critical Care ICU post delivery and received intensive chest physical therapy, midline intravenous lines and deep vein thrombosis compression devices. On admission to the unit, she required treatment for hypertension with multiple doses of intravenous Valium. On her first post-operative day, she developed apnea after a dysautonomic crisis both of which were successfully treated. After being transferred to the postpartum floor on post-operative day 2, she was noted to be febrile with a temperature of 102 degrees. The patient was treated after a fever workup for presumed aspiration with intravenous Unasyn (ampicillin and sulbactam). She was discharged on post-operative day 4 along with her baby with her pre-delivery medications in addition to Vicodin (tylenol and hydrocodone), Motrin (ibuprofen) for pain and Levoquin (levofloxacin).

## Discussion

Familial dysautonomia is an auto recessive disorder resulting in a progressive neuronal degeneration process throughout the patient's life [4-7]. Virtually all cases of this HSAN have been reported in the Ashkenazi Jewish population [1-8]. In 2001, a mutation of the IKBKAP gene on chromosome 9 had been isolated causing this condition [2, 4-7]. The combination of alacrima, absent fungiform papillae on the tongue, depressed patellar reflexes, miosis of pupils with methacholine chloride into conjunctiva sac and abnormal histamine test with a narrow areola surrounding the wheal instead of a diffuse axon flare around a central wheal in a patient of Ashkenazi Jewish extraction is virtually confirmatory of the diagnosis [4-5]. Patients with familial dysautonomia show marked reduction in nonmyelinated neuronal populations as well as reduction in small diameter myelinated axons [5-7]. Expression of the disease differs among individuals and even in the same individual at different ages [6]. This neurologic disorder is characterized by varying degrees of sensory and autonomic dysfunction [1-8]. Patients typically present with abnormal suck and feeding difficulties, episodes of vomiting, abnormal sweating, pain and temperature insensitivities along with labile blood pressure absent tearing and scoliosis [1, 3-5, 7, 8].

Pregnancy in familial dysautonomia women are at considerably high risk particularly because of abrupt changes in blood pressure [1]. Patients can exhibit both extreme hypertension to profound and rapid postural hypotension without compensatory tachycardia [1, 4, 5, 7]. Alternating hypotension and hypertension not only poses significant risk to the patient but can also be the etiology of fetal distress secondary to placental insufficiency. Once postpartum, those diagnosed with familial dysautonomia experience swallowing difficulties and frequent episodes of emesis which contribute to the

development of aspiration pneumonia. With no cure for the disease, support measures are currently all that is available to assist the patient [1, 4]. This case involved a patient with significant symptoms present throughout her pregnancy secondary to her diagnosis of familial dysautonomia Riley-Day syndrome. With aggressive management of her autonomic crises, feeding issues, anemia and preterm labor utilizing a team of multi specialists, she was able to successfully deliver a healthy infant. Her subsequent close observation post delivery initially in the Intensive Care Unit treating her autonomic crises and later on the postpartum floor addressing a suspicion for aspiration pneumonia promptly, allowed this patient the ability to be discharged home along with her newborn without additional time spent in the hospital.

### Financial Disclosure

The author does not report any potential conflicts of interest.

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