BACKGROUND

RS, a first child, was born at term following an uncomplicated maternal pregnancy, labor, and delivery. She was breast-fed. There is no family history of cystic fibrosis (CF). At age 2 weeks, RS developed a diaper rash that over the next 3 weeks spread to most of her body, despite use of topical emollients, corticosteroids, and antibiotics. At her 2-month check, she was noted to be at 50th percentile for height and 20th percentile for weight. She also was noted to have frequent loose and sometimes greasy stools. When the rash failed to improve, RS was referred to a pediatric dermatology clinic at the regional university medical center. There she was thought to have superinfected acrodermatitis enteropathica and was treated with an oral cephalosporin antibiotic and zinc. A skin biopsy was read as consistent with acrodermatitis enteropathica and the initial zinc level was 49 µg/dL (lower limit of normal 62 µg/dL). A skin scraping culture grew Staphylococcus aureus. Other laboratory tests revealed low serum total protein and albumin (3.4 g/dL and 1.5 g/dL, respectively). RS's blood count was consistent with anemia (hemoglobin and hematocrit values were 6.5 g/dL and 20.5%, respectively).

At age 3 months, when she consistently vomited the zinc and her stools became diarrheal, RS was admitted to her community hospital. On admission she was noted to have a scaly rash over most of her body and mild generalized edema. She was given intravenous fluids and albumin. RS's anemia and hypoproteinemia worsened. Her blood chemistry values were significant for elevated liver enzymes (peak aspartate aminotransferase 305 IU/L and alanine aminotransferase 264 IU/L) and blood glucose (peak 172 mg/dL). RS was transferred to the regional children's hospital 3 days after admission. On the day of transfer she became febrile to 39°C orally.

At the children's hospital she was given total parenteral hyperalimentation, albumin infusions, nasogastric feeds, and was started on broad-spectrum antibiotics after a septic workup and a chest radiograph revealed a right upper lobe infiltrate. On the second day after transfer she became hypoxic and developed respiratory distress followed by hypotensive shock and disseminated intravascular coagulation. She was intubated and ventilated, given volume and pressor support, fresh frozen plasma, packed red blood cell transfusions, and eventually required high-frequency oscillatory ventilation for adequate oxygenation. Cultures of blood, urine, and endotracheal aspirates all grew S aureus and beta hemolytic Streptococcus Group B. RS developed a right-sided pneumothorax requiring multiple chest tube drainage. Serial chest films showed development of bilateral pneumatoceles. After a stormy course, RS was eventually extubated 1 month after admission.

DIAGNOSIS AND TREATMENT

While intubated and ventilated, a sweat test was performed on RS that revealed a sweat chloride of 113 mEq/L on a collection of 190 mg of sweat. Further studies showed positive stool fat and trypsin, and low serum vitamin A, E, and D25 levels. A CF transmembrane conductance regulator genotype demonstrated that RS was heterozygous for the ∆F508 and G542X mutations consistent with a diagnosis of CF.

RS was treated with systemic corticosteroids, nebulized dornase alpha, N-acetylcysteine, albuterol, and ipratropium bromide, chest physiotherapy, pancreatic enzymes, supplemental fat-soluble vitamins and zinc, iron, antibiotics, and weaning doses of narcotics. After 3 weeks, a chest computed tomography showed 3 pneumatoceles up to 2.5 cm in diameter each in the right middle lobe, 2 in the left lingula, mild central bronchial dilatations, and bilateral basilar atelectasis. An infant pulmonary function test showed normal resistance, compliance, and functional residual capacity. A pH probe was normal. RS's rash completely resolved. She was discharged at age 4.5 months on
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room air, receiving 150 Kcal/kg/day via overnight nasogastric and daytime oral feedings, chest physio-therapy 3 times daily, bronchodilators 4 times daily, dornase alpha once daily, pancreatic enzymes, and A, D, E, and K vitamins.

RS was readmitted to her local hospital 1 week after discharge for respiratory distress; her weight had fallen; she was retracting, wheezing, and mildly hypoxemic. Given her history, she was transferred back to the children’s hospital. There she was treated with intravenous fluids, oxygen, continuous albuterol nebulization, systemic corticosteroids, magnesium sulfate, and theophylline, but she did not improve.

RS was intubated and ventilated, but this time was easily weaned within 24 hours back to nasal cannula oxygen and then room air. She was discharged on the fifth hospital day, on an oral corticosteroid taper, and her chronic regime was augmented by addition of inhaled beclomethasone and increase of dornase alpha to twice daily. That was RS's last hospital admission. Her family has faithfully brought her to quarterly CF clinic visits since then.

LONG-TERM FOLLOW-UP

RS is now 10 years old, and she was last seen for a routine quarterly visit at the CF center in the summer of 2007. At that visit, she was clinically well, having completed a stint at sleep-away camp, playing tennis, and having no complaints or symptoms related to CF. Her CF regime includes pancreatic enzymes (approximately 2000 U lipase/kg/meal) with all meals, a fat-soluble vitamin supplement, 2 puffs of an albuterol-iptropium inhaler daily in the morning before dornase alpha nebulization and a 20-minute Vest airway clearance treatment, 7% hypertonic saline nebulized twice daily, fluticasone 44 µg inhaler 2 puffs daily, budesonide nasal spray daily, docusate 100 mg daily, vigorous daily exercise, and a high-fat high-protein diet. Microbiological cultures of her oropharyngeal tract grow S aureus; she has never grown Pseudomonas aeruginosa. RS is enrolled in the Early Pseudomonas Infection Control Observational Trial and a phase III clinical trial of inhaled denufosol, an experimental ion transport modulator. RS's body mass index is in the 65th percentile, her physical examination is benign, and her spirometric lung function shows a forced expiratory volume in 1 second of 2.02 L (94% predicted). Her most recent chest radiograph in spring 2007 showed a Brasfield CF chest radiograph score of 20, and her last chest computed tomography in March 2005 revealed mild bronchiectatic changes in all lobes, particularly the right middle lobe, which also showed mucus plugging and air trapping. RS will continue to be followed at least quarterly, and is currently stable and doing well.

DISCUSSION

This case illustrates that a severe, even life-threatening, early presentation of CF can be reversed with proper therapy. The patient’s current status suggests a good prognosis based upon her lung function and imaging, nutritional status, and lack of chronic respiratory infection with P aeruginosa.