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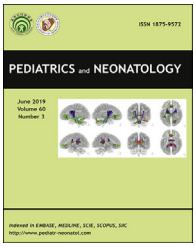

Letter to the Editor

Reply — Cystic fibrosis complicated by cor pulmonale: The first case report in Taiwan

To the Editor,

Moslehi[1] had some recommendations on our paper about "Cystic Fibrosis Complicated by Cor Pulmonale: The First Case Report in Taiwan."[2] He mentioned the possibility of neglected and/or undiagnosed severe cystic fibrosis (CF) patients in Asia and Taiwan and had some suggestions about prompting the CF condition among Asian countries and Taiwan.[1] CF is an autosomal recessive inherited disorder most commonly found in Caucasians and characterized by a defective chloride transport channel in the epithelial cells. CF is rare among Asians, and even rarer in Taiwan. We have only experienced 15 cases, including two expired siblings with similar symptoms and two patients with mixed parentage.[2–5] The gene mutations reported for these patients include 1898 + 5G → T in two patients; 1898 + 5G → T/2215insG in two patients; heterozygous G151T from the father/989-992insA from the mother in two patients (Vietnamese mother); R553X/R553X in one patient; 3849t10kb C->T in two patients; heterozygous c. 1898t5 G->T and heterozygous p. I1023R in three patients; and heterozygous delta F508 mutation from the father/13 TG repeats in the IVS8-5T from the mother in two patients (Australian father).[5] Even in the same family with the same gene mutations, our CF patients have a remarkable difference in their disease pattern and severity among siblings. Lung disease majorly causes morbidity and mortality in our CF patients. Only one patient died because of cor pulmonale with early onset and rapidly progressive symptoms.[2,3] This patient had chronic diarrhea with failure to thrive and frequent respiratory tract infections beginning at the age of 2 months. He developed bronchiectasis with chronic severe hypoxemia and pancreatic insufficiency by the age of 3 years and 5 months. He received intensive chest care and inhaled tobramycin, azithromycin, and pancreatic enzyme supplement as maintenance therapy, but his disease still progressed. CF with diffuse bronchiectasis complicated by cor pulmonale was diagnosed when he was 8 years old, and he died 2 months later despite intensive management because he was not able to undergo transplantation.[2]

In Taiwan, we have our rare disease law and rare disease foundation. CF is considered one of our official rare diseases. After a patient's diagnosis is confirmed by the national CF committee and registries, the patient can receive updated therapies including tobramycin inhalation without any payment. In Taiwan, we can arrange the sweat chloride test and genetic studies in our medical centers. We constantly bring CF awareness through media. We have tried to set up our national and regional CF frameworks, and we also tried to link them with other national and international registry systems. Because of the rarity of a patient with CF in Taiwan, we still did not set up our national or international multicenter studies and CF neonatal screening test at present.

Conflicts of interest

None declared.

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