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Title: A new mutation of surfactant protein C gene causing severe respiratory insufficiency and pulmonary fibrosis

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Body: The pulmonary surfactant is an unique phospholipid and protein complex that is synthesized, packaged and secreted by alveolar type II cells. The phospholipid compontents constitute approximately 90% weight of pulmonary surfactant, while the remaining 10% is constituted by protein components, including surfactant-associated proteins SP-A, B, C and D. Genetic mutations in surfactant production and function are associated with different clinical phenotypes and can cause significant primary lung disease in full-term infants, older children and adults. Mutations in the surfactant protein C gene have been recently associated with the development of diffuse lung disease, particularly sporadic and familial interstitial lung disease. SC is a one year old girl, who presented acute respiratory insufficiency at 6 months of age during an acute bronchiolitis, requiring intubation and mechanical ventilation. CT scan showed bilateral ground glass appearance and siderocytes were found in BAL fluid. She was referred 6 months later in poor general conditions, failure to thrive and oxygen dependent. Cystic fibrosis was excluded by negative sweat tests and CFTR mutations study. CT scan confirmed diffuse interstitial lung disease and “honeycombing” appearance. Pulmonary biopsy showed an interstitial lung disease with lamellar bodies anomalies typical of surfactant proteins deficiency. The genetic mutations analysis showed a new mutation in the exon 3 of the gene encoding SP-C.Patients with severe respiratory failure and/or interstitial lung disease should be always investigated for surfactant protein deficiency, particularly if other more common diseases are excluded.