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Immunofluorescence Analysis Results of Patients with Suspected Primary Ciliary Dyskinesia

Emine Atağ[1], Rim Hjeij[2], Nilay Bas İkizoğlu[3], Pınar Ergenekon[4], Cansu Yiğit Yılmaz[4], Yasemin Gökdemir[4], Ela Erdem Eralp[4], Heymut Omran[2], Bülent Karadağ[4]

[1]Ümraniye Training and Research Hospital, İstanbul, Turkey
[2]Division of Pediatric Pulmonology, Marmara University School of Medicine, İstanbul, Turkey
[3]Süreyyapaşa Chest Diseases and Chest Surgery Training and Research Hospital, İstanbul, Turkey
[4]Department of General Pediatrics, University Hospital Muenster, Muenster, Germany

Objectives: Primary ciliary dyskinesia (PCD) is a genetic disease characterized by impaired mucociliary function and recurrent sinopulmonary infections. High-speed video microscopy analysis and nasal nitric oxide levels were used as screening tests for diagnosis. Immunofluorescence (IF) staining of cilia proteins is gaining importance in recent years. In this study the results of immunofluorescence ana-lyses (IF) of 36 Turkish patients with suspected PCD were reported.

Methods: Thirty six patients who were followed-up at Marmara University Pediatric Pulmonology Department were included in the study. Nasal brush samples of all patients were evaluated by IF staining with antibodies against DNAH5, GAS8, DNAH11, RSPH9 proteins. In case of negativity for these IF analysis; further IF analyses were performed tackling the proteins DNALI1 and CCDC39.

Results: Mean age of patients was 12,96±0,72 years. 52% (n=19) of patients were girls. 78% of patients had a history of term birth and in 64% (n=23) of patients, the symptoms started in the neonatal period. Twenty two (61%) patients had consanguinity between parents. 91% of patient had productive cough. Chest deformity was present in 1 patient, and clubbing was present in 7 patients. Oxygen requirement was present in 19% (n=7) patients. Nineteen of the patients had dextrocardia (52%) and 16 patients (44%) had situs inversus totalis. Lung function tests results were as mean values FEV1:77.96%±16.65, FVC: 82%±14.11, FEV1/FVC: 92.78±10.02, PEF: 80.56%±18.72, FEF25-75: 68.84%±28.87. The median number of use of antibiotics was 2(1, 3(25. and 75. percentiles)) and the median number of hospitalizations was 0 [0, 0.75 (25. ve 75. percentiles)]. Comparision of ciliary defects with clinical features did not reveal significant difference in lung function tests, the number of antibiotic usage and hospitalization with IF defect type ($p>0.05$). Out of 36 patients, 12 had an outer dynein arm defect, 7 had an inner dynein arm defect, 7 had an N-DRC defect and 4 had a radial spoke defect. In 6 patients (16.6%), no defect was detected by IF examination.

Conclusion: Examination of ciliated proteins by IF analysis facilitates genetic diagnosis by analysing target genes instead of whole genome. In this study, 30 patients out of 36 could be confirmed to have the disease, emphasising the power of IF in the diagnosis of PCD. The remaining 6 individuals should be subject to further IF analyses and complementary diagnostic tests.

Keywords: Immunofluorescence analysis, paediatric, primary ciliary dyskinesia