A complete test panel avoids missed PCD diagnosis in adults with mild clinical phenotype

D. Calmés[1], N. Bricmont[2], R. Bonhiver[2], F. Guissard[1], C. Pauly[3], O. Hougrand[4], L. Benchimol[5], A.L. Poirrier[5], E. Docampo Martinez[6], J. Harvengt[6], A. Jacquinet[6], M.C. Seghaye[7], R. Louis[1,2], C. Kempeneers[2,3]

[1]: Department of Pneumology, University Hospital Liège, Belgium, [2]: I3 Group, GIGA Research Center, University Hospital Liège, Belgium, [3]: Division of Respirology, Department of Pediatrics, University Hospital Liège, Belgium, [4]: Unit of Electron Microscopy, Department of Pathology, Unilab LG, University Hospital Liège, Belgium, [5]: Department of ENT, University Hospital Liège, Belgium, [6]: Department of Genetics, University Hospital Liège, Belgium, [7]: Division of Cardiology, Department of Pediatrics, University Hospital Liège, Belgium

Introduction

Primary ciliary dyskinesia (PCD) is a heterogeneous genetic motile ciliopathy. Diagnosis is difficult as there is no single gold standard reference test.

A combination of clinical history, nasal nitric oxide (nNO), high-speed video microscopy analysis (HSVA), immunofluorescence labelling of ciliary proteins (IF), transmission electron microscopy (TEM) and genetic testing is currently used.

ERS guidelines state that PCD can be confirmed by TEM or genotyping, but this will miss the diagnosis in 15-30 % of cases.

Data in adults is scarcer than in the pediatric population.

Aim: To evaluate how PCD was diagnosed in an adult population

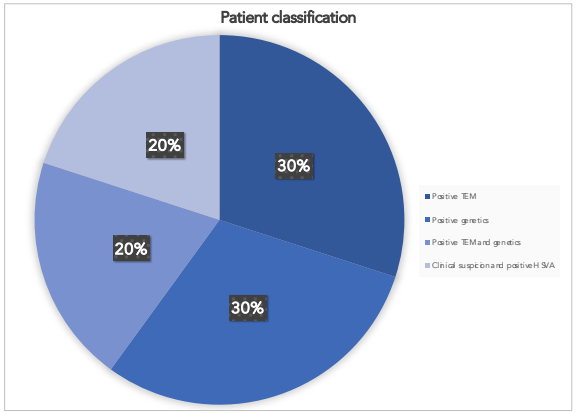
Methods

This was a retrospective study on 19 adult patients referred to our PCD diagnostic center. Most patients underwent nNO, HSVA, IF, TEM and genetic testing.

All cases were discussed in a multidisciplinary team (MDT) meeting to come to a positive, negative or inconclusive PCD diagnosis.

Results

Patients were classified based on the MDT final report: 10 PCD positive, 6 PCD negative and 3 inconclusive. Median age of PCD positive patients was 46 (20-82) years, with 2/10 having positive TEM and genetics, 3/10 positive TEM only, 3/10 positive genetics only and 2/10 normal TEM and genetics diagnosed after MDT meeting.



2 PCD positive had a low clinical suspicion (PICADAR<5). One had low nNO, abnormal HSVA, positive TEM and IF, unavailable genetics. The other had normal nNO, normal HSVA, positive TEM, unavailable IF, negative genetics.

Conclusion

Our results confirm that 20% of adult PCD cases are missed by TEM and genetics. Moreover, our series suggest that even mild phenotypes should benefit from the complete test panel, if available, to avoid missed PCD diagnosis.