

Poster Abstract: Diagnostic

Cine-MRI as a New Tool to Evaluate Diaphragmatic Dysfunction in Pompe Disease

Stephan C. Wens^{1,2}, Pierluigi Ciet^{3,4}, Adria Perez-Rovira^{3,5,6}, Karla Logie⁶, Elizabeth Salamon⁶, Piotr Wielopolski³, Marleen de Bruijne^{3,5}, Michelle E. Kruijshaar², Harm W. Tiddens^{3,4}, Nadine A.M.E. van der Beek^{1,2}, Pieter A. van Doorn^{1,2}, Ans T. van der Ploeg^{2,7,*}

¹Department of Neurology, Erasmus MC University Medical Center, Rotterdam, Netherlands

²Center for Lysosomal and Metabolic Diseases, Erasmus MC University Medical Center, Rotterdam, Netherlands

³Department of Radiology, Erasmus MC University Medical Center, Rotterdam, Netherlands

⁴Department of Pediatrics, Respiratory Medicine and Allergology, Erasmus MC – Sophia Children's Hospital, University Medical Center, Rotterdam, Netherlands

⁵Department of Biomedical Imaging Group Rotterdam, Erasmus MC University Medical Center, Rotterdam, Netherlands

⁶Department of Pediatric Pulmonology, Erasmus MC – Sophia Children's Hospital, University Medical Center, Rotterdam, Netherlands

⁷Department of Pediatrics, Division of Metabolic Diseases and Genetics, Erasmus MC – Sophia Children's Hospital, University Medical Center, Rotterdam, Netherlands

BACKGROUND

Severe pulmonary dysfunction is a serious threat to patients with Pompe disease, a treatable metabolic neuromuscular disorder caused by lysosomal acid alpha-glucosidase deficiency. This pulmonary dysfunction – which is particularly severe in the supine position – is mainly caused by diaphragmatic weakness. Standard pulmonary function tests only provide indirect information about diaphragmatic function, and they do not supply information about chest mechanics in detail. We therefore used cine-MRI to examine the dynamic performance of respiratory muscles, and compared these data with the results of simultaneously performed pulmonary function testing.

METHODS

Ten adult patients with Pompe disease and six healthy volunteers participated. We performed two static scans at end-inspiration and end-expiration to evaluate lung anatomy and lung volumes. Three dynamic 3D acquisitions were performed to investigate overall respiratory dynamics. Using manual segmen-

tation of the acquired images, three length ratios were calculated. Diaphragmatic displacement manifests itself by motion in the craniocaudal direction, while movement in anteroposterior and left–right directions reflects chest wall displacement.

RESULTS

Patients with Pompe disease have a significantly reduced craniocaudal length ratio compared with healthy volunteers ($p < 0.001$), indicating impaired diaphragmatic displacement. This ratio correlated strongly with forced vital capacity (FVC) in the supine position ($r = 0.88$), and severity of 'postural drop' ($FVC_{\text{sitting}} - FVC_{\text{supine}}$; $r = 0.89$). The difference in anteroposterior length ratio was less pronounced ($p = 0.04$), while there was no difference in left–right length ratio ($p = 0.1$).

CONCLUSIONS

Cine-MRI is a promising technique to assess chest mechanics and to visualize the severely impaired diaphragmatic function in patients with Pompe disease. It may allow us to detect respiratory weakness at an earlier stage. Early diagnosis of diaphragmatic weakness may prove important in deciding when to start enzyme treatment.

*Correspondence to: Ans T. van der Ploeg, Centre for Lysosomal and Metabolic Diseases, Department of Pediatrics, Sophia Children's Hospital, PO Box 2060, 3000 CB Rotterdam, Netherlands. E-mail: a.vanderploeg@erasmusmc.nl