A radiological classification of pulmonary involvement in Niemann-Pick type B disease

Poster No.: C-1470
Congress: ECR 2014
Type: Scientific Exhibit
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Keywords: Respiratory system, Lung, CT-High Resolution, Comparative studies, Treatment effects
DOI: 10.1594/ecr2014/C-1470

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Aims and objectives

Niemann-Pick disease type B (NPB) is a rare lysosomal storage disorder caused by acid sphingomyelinase (ASM) deficiency, leading to lipid storage particularly in the reticulum endothelial cells of the liver, spleen and lungs. Pathophysiology of pulmonary involvement is still incompletely understood (Fig. 1 on page 2 shows a proposed physiopathology scheme). However, histopathological data showed that lipid-laden macrophages accumulate in the alveolar septa, bronchial walls, and pleura, causing a progressively worsening restrictive pattern at pulmonary function testing. High resolution computed tomography (HRCT) well depicts pathologic changes in these patients, even though radiological findings are not associated with clinical manifestations.

In order to build a severity score index based both on clinical and imaging data, we propose a simple scale of HRCT changes, that takes into account the extent of the pulmonary involvement.

Images for this section:
Methods and materials

Since June 2012 to July 2013 we addressed to chest X-ray and HRCT 10 patients affected by NPB disease (age range 26 - 60 y, media 40.5 ± 14.81 y), ranging from very mild to severe clinical respiratory presentation.

All patients where admitted to X-ray examination and HRCT examination at the same time. We collected also pulmonary function tests results and all clinical informations since the diagnosis time to the present.

Results

Consistently with the results from the literature, we observed two fundamental patterns: smooth septal thickening and ground-glass opacities. These were or not associated with rare nodules (dense or ground-glass) or cysts. In our experience, these findings correlate with a worsening of PFT (pulmonary function tests, in particular DLCO).

As follows, in this poster we show 4 representative cases. Based on a simple observation of these findings we propose a radiological scale of pulmonary scale (Fig. 2 on page 5).

Case 1 (Fig. 3 on page 6).

Man, 60 years-old. Good health. Normal findings at PFT [FVC 5.14 L (125%), FEV1 4.02 L (123%), TLC 6.64 L, DLCO 27.9 ml/min]. Normal findings at walking test [590 m traveled, HR before test 64/’, HR after test 92/’, RR before test 12/’, RR after test 16/’, S02 98% during the test].

Chest X-ray and HRCT of this patient doesn't show any abnormal findings.

Case 2 (Fig. 4 on page 6, Fig. 5 on page 7).

Man, 44 years-old. Diagnosis of NPB by liver biopsy at 14 yo. At 36 yo aortic dissection with subsequently cardiosurgical intervention of aortic prothesis. PFT: mild restrictive impairment [FVC 4.47 L (82%), FEV1 3.13 L (75%), TLC 5.85 L (72%), DLCO 16.2 ml/mmHg/min (61%)]. Normal findings at walking test [651 m traveled, HR before test 58/’, HR after test 100/’, RR before test 14/’, RR after test 16/’, S02 98% during the tests].

Chest X-ray and HRCT show a dishomogeneous smooth septal thickening (arrows).
Case 3 (Fig. 6 on page 7, Fig. 7 on page 8).

Woman, 33 years-old. Diagnosis of NPB by cutaneous biopsy at 2 yo. Anamnesis of hepatomegaly and recurrent respiratory infections. PFT: mild restrictive impairment [FVC 3.2L (79%), FEV1 2.2L (70%), TLC 4.25L (72%), DLCO 16.2 ml/mmHg/min (50%)]. Minimal impairment at walking test [560 m traveled, HR before test 68‘, HR after test 107‘, RR before test 13‘, RR after test 20‘, S02 99% to 94% during test].

Chest X-ray and HRCT: arrowheads indicates scattered ground-glass opacities; arrows show smooth septal thickening (as in case 2).

Case 4 (Fig. 8 on page 8, Fig. 9 on page 9).

Woman, 26 years-old. Diagnosis of NPB at 5 yo. Significant reduced resistance to exercise. PFT: severe restrictive/obstructive impairment [FVC 2.8L (75%), FEV1 2L (69%), TLC 4L (70%), DLCO 13 ml/mmHg/min (44%)]. Abnormal findings at walking test: 450 m traveled, SO2 97% to 80% during the test.

HRCT well depicts diffuse crazy-paving pattern (apical-basal gradient) due to coexistence of smooth septal thickening and ground-glass opacities in intermixed pattern.

Unusual Findings (Fig. 10 on page 9).

In case 2 and 3, HRCT has also identified rare unusual findings, consistently with previous literature reports. We observed some low density nodules (Fig. 10 on page 9 arrow) or high density nodules (Fig. 10 on page 9 arrowhead), ranging 5 mm to 1.5 cm.

In the Fig. 10 on page 9 is represented also a cystic lesion (with a well defined wall) near the bronchial structures at the right lung ilum.

Images for this section:
Table 1. Major Findings

<table>
<thead>
<tr>
<th>Scale</th>
<th>Major Findings</th>
<th>Minor Findings</th>
</tr>
</thead>
<tbody>
<tr>
<td>I</td>
<td>Normal findings</td>
<td>None</td>
</tr>
<tr>
<td>II</td>
<td>Smooth septal thickening</td>
<td>Nodules</td>
</tr>
<tr>
<td>III</td>
<td>Smooth septal thickening + ground-glass opacities</td>
<td>Nodules</td>
</tr>
<tr>
<td>IV</td>
<td>Smooth septal thickening + ground-glass opacities in intermixed pattern (crazy-paving)</td>
<td>Nodules</td>
</tr>
</tbody>
</table>

Fig. 2

Fig. 3
Conclusion

Radiological findings are very clear but absolutely aspecific. Table 1 shows our simple radiological scale based on HRCT findings. As previously reported, these findings well correlate with modifications of PFT. This could be used both to classify patients at the time of diagnosis and to monitor the effect of specific treatments during the follow-up. Moreover, the possible role of radiological findings as predictors of clinical worsening should be investigated by a larger study. Infact, the limit of this study is low number of patients examined.

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