The significance of MRI in myelin disorders

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Systematic analysis of white matter changes on magnetic resonance (MR) images has increased the diagnostic quality of MR interpretation. For that purpose, structural elements were defined, relevant to the distinction of disease groups. Per disease group histograms were obtained of the frequency of involvement of these structural elements. On the reverse, this database could be used as the basis of an expert system. Loaded with the data of a new case, the program comes up with a differential diagnosis and probability percentage with confidence intervals per diagnostic item. The program has helped in identifying MR patterns in rare disorders.

Keywords: white matter disorders, myelin disorders, MRI of myelin disorders, pattern recognition.

Histologically distinction is possible between a selective white matter disorders and selective myelin disorders. This distinction cannot be made in most cases by magnetic resonance imaging (MRI). We will, therefore, use white matter disorders (WMD) as a heading, including myelin disorders.

MRI has high sensitivity in visualizing WMD. To improve the specificity, we developed a systemic approach to the MR analysis of WMD [1]. Structural elements were defined relevant to distinguishing between entities. A scorings list was prepared and each of 43 items was scored per patient. The obtained data were collected in a database, which made it subsequently possible to obtain histograms of the frequency of involvement of structured elements per disease group (Fig. 1). Additional characteristics, such as calcification, cyst formation, necrosis, and hemorrhage were also scored. The program could now be used as an expert system. When data of a new case were imported in the program, the computer came up with a diagnostic suggestion, a probability percentage, and confidence intervals. All cases in the database (1483) were verified histologically or by laboratory investigations.

To use a pattern recognition program one obviously needs a practical classification of the concerned WMD. We proposed such a classification in previous work [1, 2].

WMD can be subdivided into acquired and hereditary disorders. The hereditary disorders can, in our opinion, best be classified by the cellular substructure, the organelle, in which the deficient enzyme is normally present. Accordingly, we distinguish lysosomal, peroxosomal, mitochondrial, and cytoplasmic disorders, disorders with a nuclear enzyme defect, and disorders of unknown etiology.

In the category of organic acid and amino acid metabolism disorders, the locations of the defect are not uniform and are not always known. This group is, therefore, treated as a separate entity.

The acquired disorders are clustered as inflammato-ry–infectious, toxic–metabolic, hypoxic–ischemic, and traumatic WMD. This classification serves its purpose for the pattern-recognition program and leaves enough space to integrate new findings.

LYSOMAL STORAGE DISORDERS

In lysosomal storage disorders, specific catabolic enzymes are absent and this causes the accumulation of metabolites. Four major categories of lysosomal storage disorders can be distinguished: the sphingolipido-
Fig. 1. Computer histograms of the frequency of involvement of structural elements in X-linked adrenoleukodystrophy and Pelizaeus–Merzbacher’s disease.

Fig. 2. Transverse $T_1$-weighted (a) and $T_2$-weighted (b) images in a patient with metachromatic leukodystrophy. Periventricular area with high signal intensity on the $T_2$-weighted image. The arcuate fibers are spared in the beginning [arrow in (a)].

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