Recognizable phenotypes associated with intracranial calcification.

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Abstract

AIM:

In this observational study, we adopted a systematic approach to the radiological phenotyping of disorders associated with intracranial calcification, with the aim of determining if characteristic patterns could be defined as an aid to the future diagnosis of known conditions and the identification of new disorders.

METHOD:

A cranial imaging-based scoring system was devised using both computed tomography and magnetic resonance imaging data. Patients were grouped into diagnostic categories where a definitive molecular diagnosis was known, or where the clinical and radiological features suggested a specific diagnosis. For patients in whom the diagnosis was unknown, subgroups were defined according to shared radiological features.

RESULTS:

Data on 244 scans from 119 patients were analysed. A specific diagnosis was available for 59 patients (31 males, 28 females; median age 50 mo, range 1 wk to 54 y). These were as follows (number of patients in brackets): Aicardi-Goutières syndrome (33), cerebroretinal microangiopathy with calcification and cysts (10), band-like calcification with simplified gyration and polymicrogyria (6), COL4A1-related disease (3), Degas disease (2), Krabbe disease (2), Alexander disease (1), mitochondrial disease (1), and tetrasomy 15 (1). In 60 patients the aetiology was unknown. Within this group, subsets demonstrating shared characteristics suggestive of a specific calcification phenotype could be identified.

INTERPRETATION:

This study confirms the value of a systematic approach to radiological phenotyping of disorders associated with intracranial calcification.

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