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Autosomal recessive transmission of chorea-acanthocytosis confirmed

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We would like to draw attention to new genetic data recently reported [8] on a patient previously described as suffering from chorea-acanthocytosis (ChAc) with autosomal dominant transmission [7] and for whom neuropathological findings were published in this journal in 2009 [4]. At that time we questioned the apparent inheritance, given the limitations of the molecular methodology employed, and the lack of support for this mode of transmission otherwise found [2].

A correction concerning the original publication by Saiki et al. [7] has recently been published [1] stating: “an error in sequencing occurred and the inheritance pattern should have been reported as autosomal recessive (pseudodominant)”. This erratum reflects the findings reported by Tomiyasu et al. [8] whose patient no. 22 was identified with III-3 from the pedigree originally published and is identical with the patient in whom autopsy findings had been described in this journal [4]. This subject was also reported as case 1 of a further clinical study [6]. The new molecular findings describe compound heterozygous *VPS13A* mutations in him and also in his affected sister: 1305G>A in exon 15 and 8035G>A in exon 57. The nomenclature of the latter mutation was corrected from 8295G>A in the original report [4, 7]. The patients’ mother (II-4) was shown to be an asymptomatic carrier of a single heterozygous 1305G>A mutation.

The father of the postmortem case of Ishida et al. [4], by conclusion a carrier of the 8035G>A mutation, was said to be suffering from early onset hyperkinesia as did other family members [5]. Unfortunately, no further analyses on DNA were feasible and the presence of mutations in other genes, e.g., *IT15*, responsible for the supposedly dominant features observed in this man, his father, his two sisters and a sister’s son could not be investigated [7]. In addition, there is still a possibility of subclinical involvement in subjects who carry a single heterozygous *VPS13A* mutation although, so far, no convincing evidence has yet been put forward to support that claim [3, 9].

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In summary, the suggestion of dominant transmission of a condition recognized as an autosomal recessive trait (OMIM 200150) has eventually been put to rest in the autopsy case reported in this journal in 2009 and one source of confusion has been removed.

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