that gender was not an important confounder. The prevalence of the I allele in the population was 0.4, similar to that reported in the Jamaican black population.[7 8] The sample population is in Hardy-Weinberg equilibrium.

[1]Three reports on black populations have been published, one on American blacks which showed an absence of any association between genotype and ACE serum levels,[6] while the others showed a significant effect of the D allele.[7 8] These last two studies, both on Jamaican populations, suggested the same general trend but with slightly differing results. Forrester *et al*[7] showed a significant difference between serum ACE in all three genotypic groups with the same codominant effect seen in whites. McKenzie *et al*[8] also showed significance between all groups, but this was less pronounced between II and ID than for any of the other cross comparisons. The ACE levels reported by McKenzie *et al*[8] showed considerable overlap between groups, and this was especially prominent between the ID and DD groups. We also found considerable variation in the ACE serum levels within genotypic groups, with the greatest scatter in the ID and DD groups. There were significantly lower serum ACE levels in the II group compared to the ID or DD groups in the Ghanaian population, but no difference between the ID or DD groups. This trend is seen in the data of McKenzie *et al*,[8] but not at all in that of Forrester *et al*.[7] Since these two reports studied the same population, it is possible that the difference is simply a statistical artefact.

Our data show that in a black African population the trend in McKenzie *et al*[8] is increased to produce a dominant effect of the D allele on ACE serum levels rather than codominant. It may be that there has been genetic input from white gene pools in the Jamaican population which has produced a less dominant relationship between the *ACE* D allele and serum ACE levels than we have shown. The fact that Forrester *et al*[7] attempted to show black ethnicity by having "three or four grandparents of predominantly African origin" shows the problems with such a population. Unlike Blom *et al*,[6] we do find a relationship between ACE I/D polymorphism and ACE serum levels in the black population, but one where the D allele shows dominance rather that codominance. The numbers in this study are not large and the data could be influenced by this, but the sample is larger than that used by Rigat *et al*[1] to show the codominant influence of the I and D alleles in whites. Nonetheless, a much larger study in this or another black African population would be useful to confirm these data, with a matched white population as a comparison.

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Coexistence of Gaucher disease type 1 and Joubert syndrome

EDITOR—Van Royen-Kerkhof *et al*[1] reported two boys with Gaucher disease type 1 and Joubert syndrome (JS). Their case 1 had, in addition to mental retardation, choreoretinal colobomas, cerebellar vermis agenesis, and abnormal breathing, agenesis of the corpus callosum, hydrocephalus (no further details given), and generalised seizures. Their case 2 had prenatal hydrocephalus and "fulfilled the diagnostic criteria for JS". Information about brain anatomy and retinal findings in case 2 is lacking.

We question the diagnosis of JS in these two patients. The authors cite a 1992 paper[2] but fail to reference 1997 and 1998 publications that better define the phenotype and characteristic neuroimaging of JS.[3–5] In these most recent publications, the "molar tooth sign" is defined as well as a number of distinct posterior fossa abnormalities not discussed by Van Royen-Kerkhof *et al*.[1] This is a significant omission because vermis hypoplasia alone is not pathognomonic for JS and can be seen in mimicking conditions that produce congenital oculomotor apraxia and ataxia.[6 7] In addition, to the best of our knowledge, corpus callosum agenesis, hydrocephalus, and generalised seizures are not associated with JS, as these features were not encountered in our combined series of more than 60 JS patients. At a 1998 Child Neurology Society symposium on JS sponsored by the National Institutes of Health, there was general agreement about the common and occasional abnormalities in JS that did not include the clinical or radiological features reported by Van Royen-Kerkhof *et al*[1] in the two children with Gaucher disease type 1. The proceedings of the symposium will appear in the *Journal of Child Neurology* in the autumn of 1999.

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This letter was shown to Drs van Royen-Kerkhof *et al*, who reply as follows.

We thank Drs Boltshauser and Maria for their comments on our Letter to the Editor.[1] The diagnosis of Joubert syndrome (JS) in case 1 was made 17 years ago on clinical grounds including a CT scan; MRI scanning was not yet available in those days. We were not aware of the paper of 1997.[3] The other papers the authors mention had not been published at the time our paper was submitted. As a consequence the "molar tooth sign" was not discussed. We think that corpus callosum agenesis and hydrocephalus are rather non-specific developmental defects whose presence or absence do not argue significantly for or against JS. In our opinion the presence in both cases 1 and 4 of episodic hyperpnoea/apnoea remains an argument in favour of JS.

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