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Commentary on: Margolis-Nunno H, Brenner L, Cascardi J, Koblinsky L. A new allele of the short tandem repeat (STR) locus, CSF1PO. J Forensic Sci 2001:46(6):1480–1483.

Sir:

As a user of short tandem repeat (STR) systems that include the CSF1PO locus (Geneprint CTT Multiplex and Geneprint Power-Plex® 16 System, Promega Corporation, Madison, WI), I was surprised at the authors assertion that the detection of a CSF1PO₁₆ allele represented the finding of a "previously unreported size variant." A truncated literature search revealed three previous publications of population data that include this allele (1-3). Rare alleles, as mentioned by the authors, "could have great significance in human identification." Similarly, an internationally refereed publication claiming unfounded novelty for a particular allele, in this case, the CSF1PO16, could also have great significance in influencing a scientist's conclusions in a case involving this particular genotype. I applaud the clarification undertaken by the authors as to the sequence of this rare allele and agree that this test of authenticity is required. I find it highly ironic, however, that a very simple and fundamental review of the literature refutes the primary claim of this publication that is represented in the title, "A New Allele of the Short Tandem Repeat (STR) Locus, CSF1PO."

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Author's Response

Sir:

We would like to thank Simon J. Walsh for bringing to our attention the population genetics studies (1–3) in which rare findings of a putative 16 repeat CSF1PO allele (frequencies of 0.002(1), 0.0024(2), 1 in 10,000(3)) were mentioned. We regret that our search of the literature for off-ladder CSF1PO alleles using major databases failed to reveal that such an allele had been previously referred to. In December of 1999, our 16 repeat allele was posted on the STR Base (www.cstl.nist.gov/div831/strbase) as a sequenced, unpublished variant of CSF1PO.

It has been shown that for unsequenced off-ladder or variant alleles, size alone does not confirm the existence of a new allele. A putative 15 repeat allele of D1S80 which was produced using D1S80 primers was shown by sequence analysis to be a pseudoallele and not a true 15 repeat (4). Thus, the authenticity of a new allele *can only* be established by sequencing.

Thus, although a putative 16 repeat CSF1PO allele has been previously cited in population studies as rare, ours is the first confirmed report that this allele exists.

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