Mutations in LPIN1 Cause Recurrent Acute Myoglobinuria in Childhood

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Table 1 contains errors in the "Genotype" column. The mutation of patient L.R. should be described as $c.2401C \rightarrow T$, R801X. The mutation of patient D.A. should be described as $c.192+2T \rightarrow C$ leading to the deletion of the last 106 nt of exon 2, and the mutation of patient P.N. should be described as $c.1441+2T \rightarrow C$ leading to the skipping of exon 9 (exon and nucleotide numbering according to NM_145693). A corrected version of the table is included below. The authors regret these errors.

Table 1. The Clinical, Biochemical, and Molecular Findings in the Index Patients and in an Additional Four Patients Who Were Identified by Screening of a Cohort of 22 Unrelated Patients for Mutations in the *LPIN1* Gene

| Patient (Age at Time of Writing) | Age at First Episode (Number of Episodes) | Peak CK Level (u/liter) | Family History | Muscle Findings | Genotype (Mutation at the DNA Level) | Fibroblasts' mRNA |
|--|--|-------------------------------|--|--------------------------------|--|----------------------|
| 2120, 2257, 2572 (8–10 years) | 2–7 years (1–5) | 180,000- 450,000 | Consanguineous, Arab-Muslim, seven healthy siblings | Normal lipid content | E215X/ E215X (c.643G→T) | 6% |
| 2714 (5 years) | 3 years (2) | 20,000 | Consanguineous, Palestinian-Muslim, three healthy siblings | Normal lipid content | R388X/ R388X (c.1162C→T) | not available |
| L.R. (4 years) | 15 months (6) | 100,000 | French, nonconsanguineous | moderate lipid accumulation | R801X (c.2401C \rightarrow T)/ genomic deletion of exon 18 and 19 ^a | 44% |
| D.A. (5 years) | 27 months (3) | 200,000 | Mauritanien, consanguineous | moderate lipid accumulation | c.192+2T→C / c.192+2T→C ^b | 74% |
| P.N. (8 years) | 18 months (4) | 80,000 | French, nonconsanguineous. A brother died of rhabdomyolysis at 16 months | not available | c.1441+2T \rightarrow C ^c / genomic deletion of exon 18 and 19 ^a | Not available |

^a Patients L.R. and P.N. were heterozygous for a ~2 Kb genomic deletion that included exon 18 and 19. The borders of this deletion were not determined.

^b Patient D.A. was homozygous for a $c.192+2T \rightarrow C$ mutation that resulted in the deletion of the last 106 nt of exon 2.

^c Patient P.N. was heterozygous for a c.1441+2T \rightarrow C mutation that resulted in skipping of exon 9.

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