**Additional file**

Functional influence of mutations in LYST gene was predicted by human splicing finder (HSF, version 3.1. <http://www.umd.be/HSF3>). The novel missense mutation c.5719A>G was predicted to be potential alteration of splicing. The known SNP rs201382097 (*LYST*:c.4863-4G>A) has probably no impact on splicing. However, mutations in adjacent sites of c.4863-4G could be most probably affecting splicing, indicating a potential splice site around the position.

