

**Specific gene capture combined with restriction-fragment release  
for directly fluorescent genotyping of single-nucleotide  
polymorphisms in diagnosing spinal muscular atrophy**

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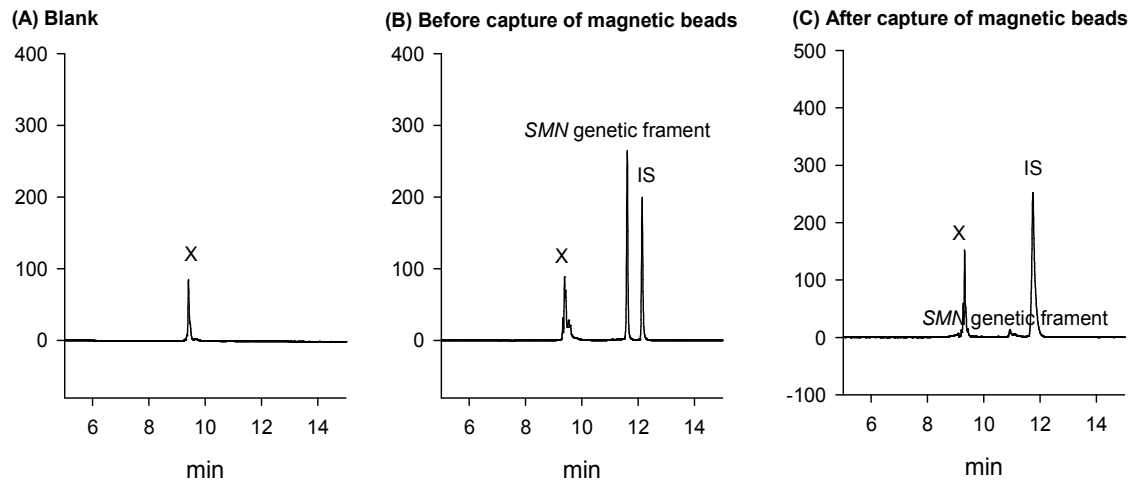


Fig. S1. The electropherograms of the PCR products of (A) blank (no genomic DNA), (B) the sample (with genomic DNA) without pretreatment of streptavidin magnet beads and (C) the sample (with genomic DNA) after pretreatment of streptavidin magnet beads. The length of *SMN* genetic fragment was 109 bp. The length of IS fragment (*KRIT1* gene) was 327 bp. The CE conditions was as followed: the separation matrix, 0.6% PEO (MW:8,000,000) in 2.0× TBE buffer; sample injection, -10 kV for 30 s; separation, -8 kV; capillary temperature, 15°C.

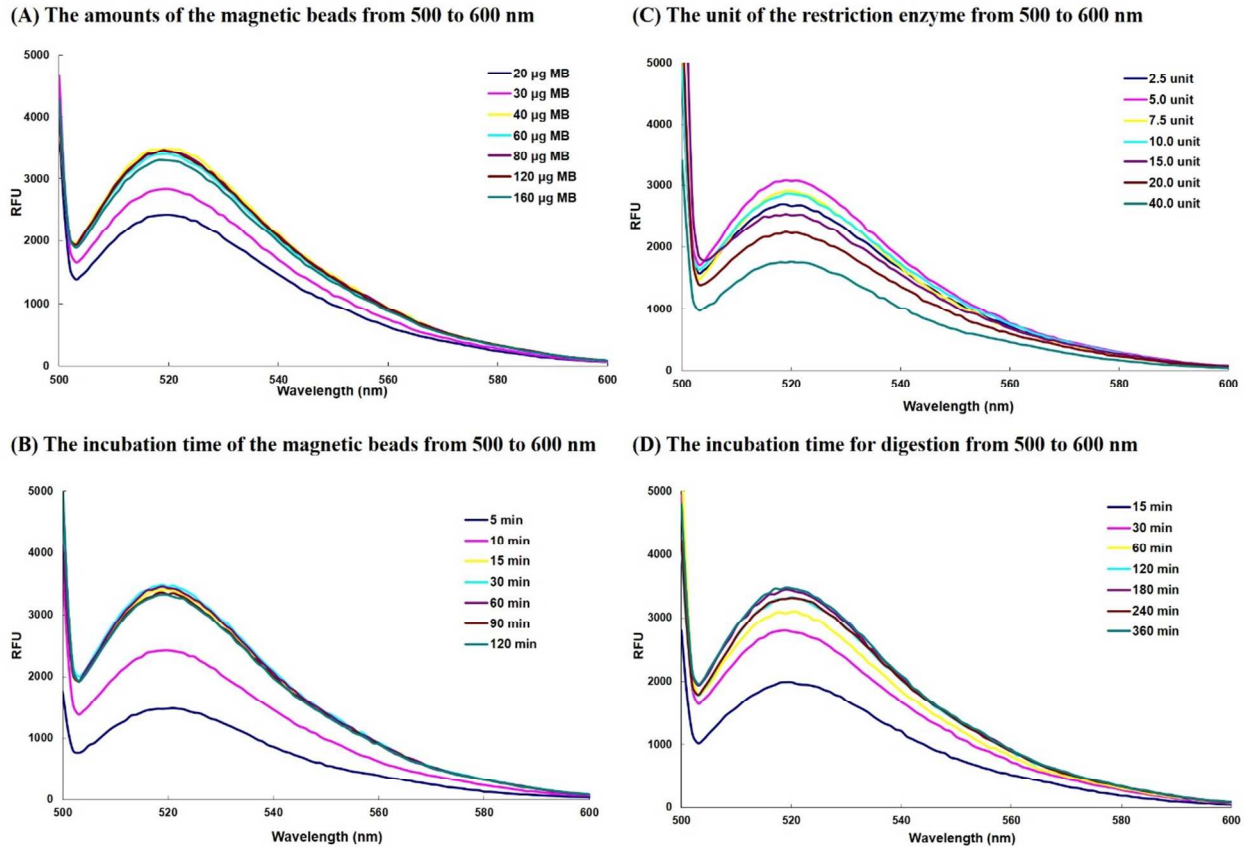


Figure S2. The effects of (A) the amount of streptavidin-coated magnetic beads, (B) incubation time of binding with biotin-coupled genetic fragments for the fluorescent determination of *SMN1* gene, (C) the amount of restriction enzyme, HPY188I, and (D) incubation time of digestion for the fluorescent quantification of *SMN1* gene. The sample was a wild control having the *SMN1/SMN2* ratio of 2/2. The results showed the various fluorescent spectra from 500 nm to 600 nm among the different conditions. The other conditions were as followed: 25 µL of the product of the biotin-coupled PCR, 5 unit of the restriction enzyme (HPY188I) and incubation time of 120 min for digestion.

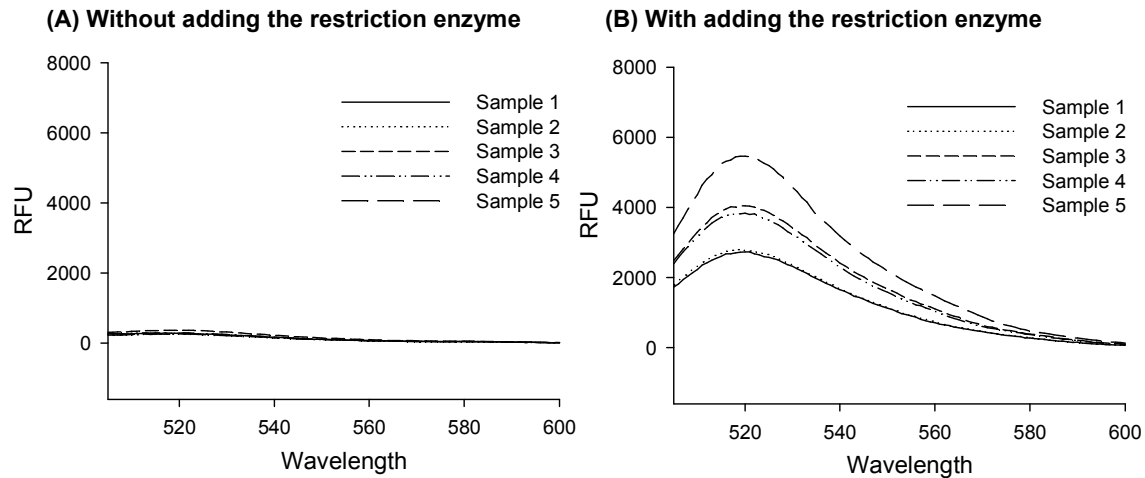


Figure S3. The effects of (A) without adding restriction enzyme and (B) with adding restriction enzyme for the digestion of *SMNI* gene in 5 real samples by fluorescent determination. The 5 clinical samples were 2 carriers having one copy of *SMNI* gene, 2 wild individuals possessing two copy of *SMNI* gene, and 1 wild subject having three copy of *SMNI* gene. The fluorescent sensitivity could be detected with adding the restriction enzyme (HPY188I) for digestion of *SMNI* gene (B), but the fluorescent sensitivity became very low without adding the restriction enzyme (A). The other conditions were as followed: 25  $\mu$ L of the product of the biotin-coupled PCR, 5 unit of the restriction enzyme (HPY188I) and incubation time of 120 min for digestion