Supplementary material to article by E. Uusitalo et al. "Neurofibromatosis Type 1 Gene Mutation Analysis Using Sequence Capture and High-throughput Sequencing"

**Fig. S1.** Pipeline for neurofibromatosis type 1 (NF1) mutation analysis. Quality of the isolated saliva DNA is shown in electrophoretic analysis on agarose gel.

1. **Saliva sampling**
   - At patient’s home using Oragene kit

2. **DNA Isolation, purification,**
   - Quality and quantity control

3. **Preparation of sample libraries**
   - Ligation of indexing adaptors, quality control

4. **Pooling of the samples**

5. **Sequence capture**
   - Protocol optimized for small target

6. **Sequencing**
   - Pyrosequencing using Roche 454GS Junior instrument

7. **Data-analysis**
   - Mapping of the reads to human genome
   - Variant calling
   - Variant filtering according to criteria

8. **Complementary tests**
   - Sanger sequencing
   - Confirmation of putative mutations and false positives
   - Sequencing of the targets with low coverage
   - MLPA
   - Copy number analysis to detect NF1 microdeletions

10 kb