

Supplemental Data

Terminal Osseous Dysplasia Is Caused by a  
Single Recurrent Mutation in the *FLNA* Gene

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Table S1. The Overview of the Data Generated by GAI

|                 | 1II:4      | 2III:5     |
|-----------------|------------|------------|
| Run             | Paired-end | Paired-end |
| Total reads     | 36,010,190 | 28,960,586 |
| Read F          | 18,005,095 | 14,480,293 |
| Read R          | 18,005,095 | 14,480,293 |
| Aligned reads   | 33,054,043 | 18,948,705 |
| Aligned in pair | 30,018,244 | 11,012,526 |
| Read length     | 51         | 51         |

Table S2. *FLNA* Primer List

| location   |                                | Primer sequences (5'-3')                         | Size (bp)                                 |
|------------|--------------------------------|--|---|
| Exon 31-32 | DNA (blood, buccal cells)      | F:GTCATCTGTGTGCGCTTTGG<br>R:AGCTGCTGAGACCGTAGAGG | 222                                       |
| Exon 31    | DNA (paraffin embedded tissue) | F:GGGCAAATACGTCATCTGTGT<br>R:agacaccctgctgacctac | 104                                       |
| Exon 29-32 | RNA                            | F:CCTGGGCGTAGGTGTACTGT<br>R:CATCAAGTACGGTGGTGACG | 416 (short isoform)<br>440 (long isoform) |
| Exon 35-37 | DNA, RNA                       | F:ACATACGCATGGAGTCGTCA<br>R:TCAACTGTGGCCATGTCCT  | 577 (DNA)<br>294 (RNA)                    |

**Figure S3. The 2100 Bioanalyzer Traces of RT-PCR on c.5217G>A from Lane1 to 8**

The peak around 15bp is the lower ladder and the signal round 1500bp is the upper ladder.

