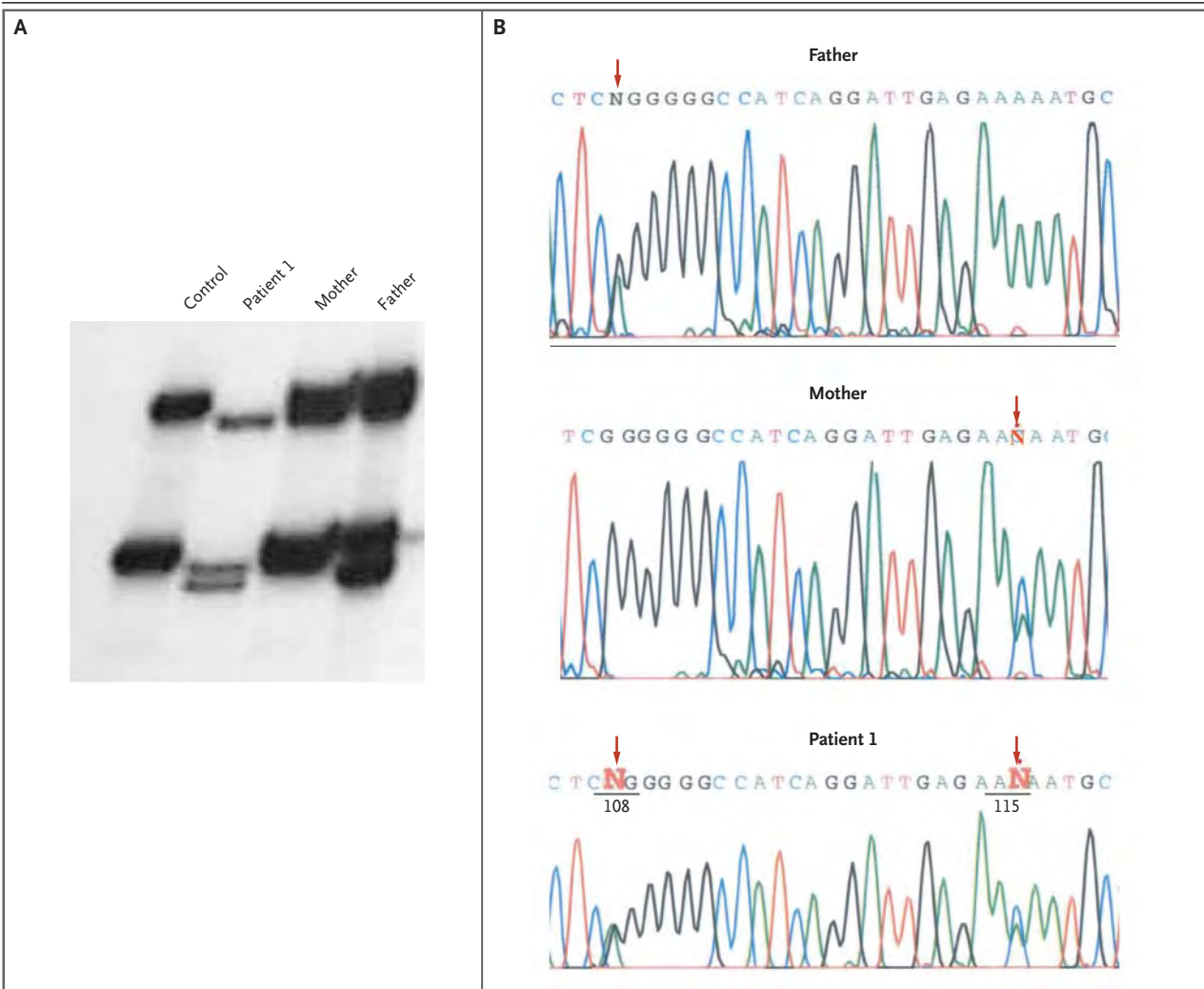


# Supplementary Appendix

This appendix has been provided by the authors to give readers additional information about their work.

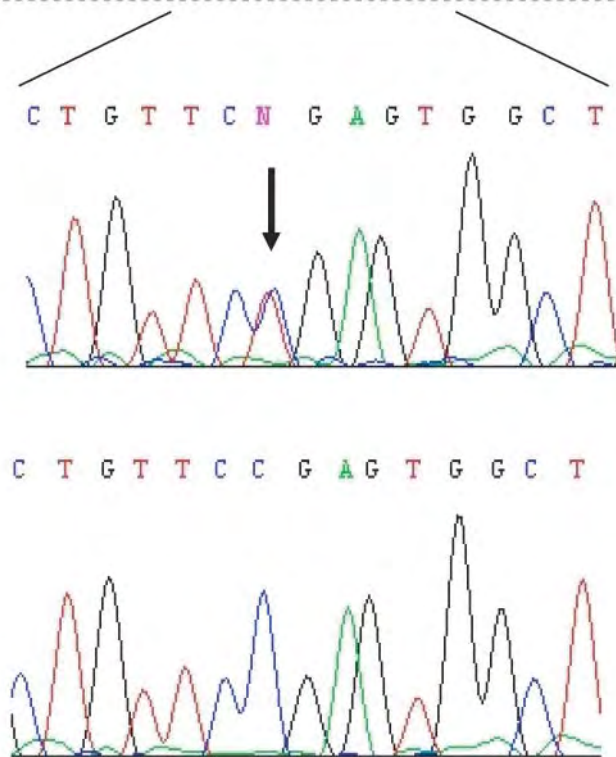
Supplement to: Abuzzahab MJ, Schneider A, Goddard A, et al. IGF-I Receptor Mutations Resulting in Intrauterine and Postnatal Growth Retardation. *N Engl J Med* 2003;349:2211-22.



**Supplementary Appendix 1. Single-Strand Conformation Polymorphism Analysis for the Middle Third of Exon 2 of the Gene for the Insulin-like Growth Factor I Receptor (*IGF-IR*) (Panel A) and the Sequence of Exon 2 DNA (Panel B) in Patient 1 and Her Parents.**

Panel A shows the normal (control) band pattern for this segment. Patient 1 has only aberrant bands. Her mother has two normal bands and the same lower aberrant band as Patient 1. Her father has the two upper aberrant bands in addition to the two normal bands. Polymerase chain reaction (PCR) used [<sup>32</sup>P]deoxycytidine triphosphate for labeling, 25 cycles with *Taq* polymerase (Life Technologies), and 5'GGACTACCGCAGCTACCGCTCC3' as the forward primer and 5'CCCACAATGTAGTTATTGGACAC3' as the reverse primer. PCR products were electrophoresed on a 0.5× mutation-detection gel (MDE, FMC Bioproducts) containing 0.6× TRIS buffer (a 1× solution that consists of 89 mM TRIS, 89 mM boric acid, and 2 mM EDTA) at 12 to 16 W in a closed-buffer-backed system for 15 to 24 hours. An autoradiogram from an MDE gel with 10 percent glycerol is shown. In Panel B, the base substitutions are indicated by arrows, and the mutated codons are underlined. The double peaks indicate heterozygosity at these loci and show that the patient is a compound heterozygote, having inherited one mutation from each parent. Sequencing of DNA was performed with the use of standard, automated methods.

1 Met Lys Ser Gly Ser Gly Gly Gly Ser Pro Thr Ser Leu Trp Gly Leu Leu Phe Leu Ser  
 1 ATGAAGTCTG GCTCCGGAGG AGGGTCCCCG ACCTCGCTGT GGGGGCTCCT GTTTCTCTCC  
 21 Ala Ala Leu Ser Leu Trp Pro Thr Ser Gly Glu Ile Cys Gly Pro Gly Ile Asp Ile Arg  
 61 GCCGCGCTCT CGCTCTGGCC GACGAGTGGG GAAATCTGCG GGCCAGGCAT CGACATCCGC  
 41 Asn Asp Tyr Gln Gln Leu Lys Arg Leu Glu Asn Cys Thr Val Ile Glu Gly Tyr Leu His  
 121 AACGACTATC AGCAGCTGAA GCGCCTGGAG AACTGCACGG TGATCGAGGG CTACCTCCAC  
 61 Ile Leu Leu Ile Ser Lys Ala Glu Asp Tyr Arg Ser Tyr Arg Phe Pro Lys Leu Thr Val  
 181 ATCCTGCTCA TCTCCAAGGC CGAGGACTAC CGCAGCTACC GCTTCCCCAA GCTCACGGTC  
 81 Ile Thr Glu Tyr Leu Leu Leu Phe <sup>Ddel</sup> Val Ala Gly Leu Glu Ser Leu Gly Asp Leu Phe  
 241 ATTACCGAGT ACTTGCTGCT GTTCTGAGTG GCTGGCCTCG AGAGCCTCGG AGACCTCTTC



**Supplementary Appendix 2. Mutation in the Gene for the Insulin-like Growth Factor I Receptor in Patient 2.**

The mutation introduces a novel restriction site (*Ddel*). Sequencing of DNA was performed with the use of standard, automated methods after polymerase chain reaction.