APPENDICES/SUPPLEMENTAL DATA

Fig 1) Array CGH profiles

Fig 1-1) Array CGH profile of patient III-2 (Family A) showing a 700 kb deletion in 11q24.3

Fig 1-2) Array CGH profile of patient III-2 (Family B) showing a 1.5Mb deletion in 11q24.2q24.3
Fig 2-1) Family A FISH analysis.
a) patient III-2, child b) patient II-3, mother
c) patient I-3; grand-father d) patient II-2, father.
Red probe: RP11-754N12 for 11q24.3 region; green probe: control probe CTC-908H22 on chromosome 11.
See the missing red probe in child, mother and grandfather analysis showing their 11q24.3 heterozygosity.

Fig 2-2) Family B FISH analysis.
a) Patient III-2, child b) Patient II-2, mother
Green probe: RP11-138K22 in 11q24.3 region
Red probe: control probe RP11-243M7 in 11p15.4.
See the unique green probe in child and mother analysis meaning that they are carrier for the 11q24.2-q24.3 deletion