

Supporting Information

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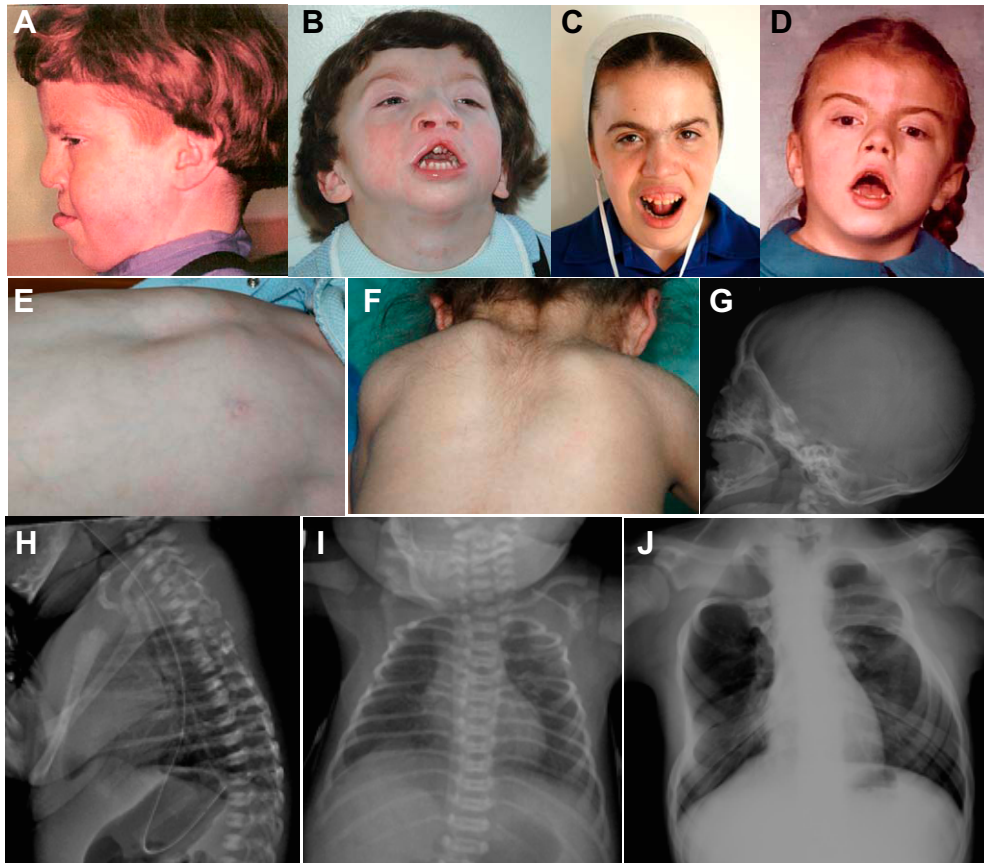


Fig. S1. Additional clinical characteristics of *TMCO1* defect syndrome. (A–D) Facial features of the syndrome in four additional patients (low hairline, brachycephaly, flat face, highly arched bushy eyebrows, synophrys, long eyelashes, orbital hypertelorism, wide nose bridge, short nose with antverted nares, microdontism, and generalized gingival hyperplasia). Parental consent has been obtained for publication of these photographs. Skeletal dysmorphisms shown here include pectus excavatum (E), Sprengel deformity of scapula (F), presence of wormian bones in the skull of a 5-day-old female (G), fusion of cervical and thoracic spine (C7 through T5 in this case) (H), and multiple rib anomalies in two different patients (I and J).

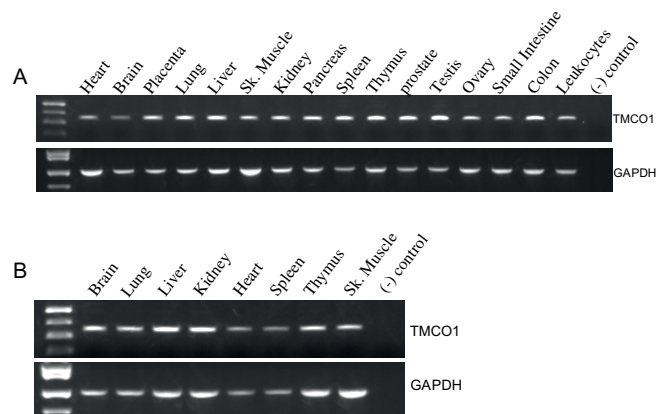


Fig. S2. Tissue expression patterns of *TMCO1* mRNA. Human multiple tissue cDNA (MTC) panels were employed as templates in PCRs containing *TMCO1* cDNA primers. A 983-bp *GAPDH* fragment was also amplified as a control of the cDNA abundance in each sample. (A) Expression in the cDNA libraries from adult human tissues. (B) Expression in fetal tissues.

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Human      MSTMFADTLLIVFISVCTALLAEGITWVLYRTDKYKRLKAEVEKQSKKLEKKKETITES 60
Orangutan  MSTMFADTLLIVFISVCTALLAEGITWVLYRTDKYKRLKAEVEKQSKKLEKKKETITES 60
Dog        MSTMFADTLLIVFISVCTALLAEGITWVLYRTDKYKRLKAEVEKQSKKLEKKKETITES 60
Horse     MSTMFADTLLIVFISVCTALLAEGITWVLYRTDKYKRLKAEVEKQSKKLEKKKETITES 60
Pig       MSTMFADTLLIVFISVCTALLAEGITWVLYRTDKYKRLKAEVEKQSKKLEKKKETITES 60
Cow       MSTMFADTLLIVFISVCTALLAEGITWVLYRTDKYKRLKAEVEKQSKKLEKKKETITES 60
Mouse     MSTMFADTLLIVFISVCTALLAEGITWVLYRTDKYKRLKAEVEKQSKKLEKKKETITES 60
Rat       MSTMFADTLLIVFISVCTALLAEGITWVLYRTDKYKRLKAEVEKQSKKLEKKKETITES 60
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Human      AGRQQKKKIERQEELKNNNRDLSMVRMKSMAIGFCFTALMGMFNSIFDGRVVAKL PFT 120
Orangutan  AGRQQKKKIERQEELKNNNRDLSMVRMKSMAIGFCFTALMGMFNSIFDGRVVAKL PFT 120
Dog        AGRQQKKKIERQEELKNNNRDLSMVRMKSMAIGFCFTALMGMFNSIFDGRVVAKL PFT 120
Horse     AGRQQKKKIERQEELKNNNRDLSMVRMKSMAIGFCFTALMGMFNSIFDGRVVAKL PFT 120
Pig       AGRQQKKKIERQEELKNNNRDLSMVRMKSMAIGFCFTALMGMFNSIFDGRVVAKL PFT 120
Cow       AGRQQKKKIERQEELKNNNRDLSMVRMKSMAIGFCFTALMGMFNSIFDGRVVAKL PFT 120
Mouse     AGRQQKKKIERQEELKNNNRDLSMVRMKSMAIGFCFTALMGMFNSIFDGRVVAKL PFT 120
Rat       AGRQQKKKIERQEELKNNNRDLSMVRMKSMAIGFCFTALMGMFNSIFDGRVVAKL PFT 120
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Human      PLSYIQGLSHRNLLGDDTTDCSFI FLYILCTMSIRQNIQKILGLAPSRAATKQAGGFLGP 180
Orangutan  PLSYIQGLSHRNLLGDDTTDCSFI FLYILCTMSIRQNIQKILGLAPSRAATKQAGGFLGP 180
Dog        PLSYIQGLSHRNLLGDDTTDCSFI FLYILCTMSIRQNIQKILGLAPSRAATKQAGGFLGP 180
Horse     PLSYIQGLSHRNLLGDDTTDCSFI FLYILCTMSIRQNIQKILGLAPSRAATKQAGGFLGP 180
Pig       PLSYIQGLSHRNLLGDDTTDCSFI FLYILCTMSIRQNIQKILGLAPSRAATKQAGGFLGP 180
Cow       PLSYIQGLSHRNLLGDDTTDCSFI FLYILCTMSIRQNIQKILGLAPSRAATKQAGGFLGP 180
Mouse     PLSYIQGLSHRNLLGDDTTDCSFI FLYILCTMSIRQNIQKILGLAPSRAATKQAGGFLGP 180
Rat       PLSYIQGLSHRNLLGDDTTDCSFI FLYILCTMSIRQNIQKILGLAPSRAATKQAGGFLGP 180
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Human      PPPSGKFS 188
Orangutan  PPPSGKFS 188
Dog        PPPSGKFS 188
Horse     PPPSGKFS 188
Pig       PPPSGKFS 188
Cow       PPPSGKFS 188
Mouse     PPPSGKFS 188
Rat       PPPSGKFS 188
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Fig. S3. ClustalW alignment of deduced amino acid sequences from eight mammalian orthologs. Identical amino acids are indicated by asterisks. Numbers on right indicate sizes.

Table S1. Primer sequences used for amplification and sequence analysis of *TMCO1*

Exon	Forward primer sequence (5'→3')	Reverse primer sequence (5'→3')	Amplicon size, bp
Genomic DNA primers for mutation screening			
1~2	GTTCTCCGATACGAGCCTTTC	CCCCTCACTCCCTCTAGTTG	917
3	TTGCAAGCTTCTTTCATTGG	GCCAAGTCAAAAGCCACA	419
4	TGCTCTGCTGCATTTGAATC	ACTTCCATTTGGTCCAGGAA	379
5	GGCTAACACGGTGAAACTCC	CCCAAAAAGTTCATGCTAAC	441
6	CATCACCGTACATCAGGTTCA	TGAGCAACTGAAAGAACTCAGG	431
7	TTCCCTTTGCCTGAGAGCTA	CTGGCTGCCATTTTCACTCT	347
cDNA primers for RT-PCR			
1~6	CGGACACTCTCCTCATCGTT	GGGGTAAAAGGAAGCTTTGC	346

Exon number corresponds to GenBank transcript accession number NM_019026. PCR conditions are available upon request.