FOXP2 variants in fourteen individuals with developmental speech and language disorders broaden the mutational and clinical spectrum

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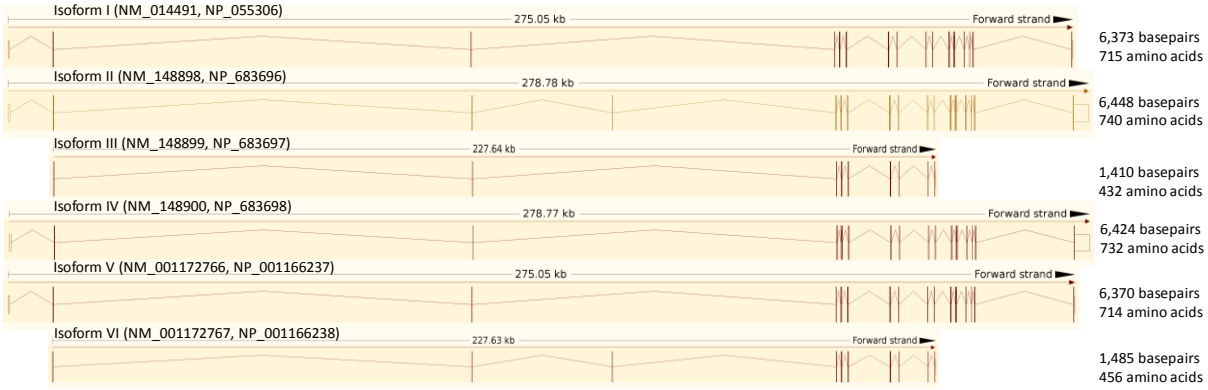
Supplementary table S1: Annotations of mutations in the four longest isoforms of FOXP2

Genomic position	Isoform II	Isoform I	Isoform IV	Isoform V
	NM_148898.3	**NM_014491.3**	**NM_148900.3**	**NM_001172766.2**
chr7:114282671C>T	c.1057C>T, p.(Arg353*)	c.982C>T, p.(Arg328*)	c.1033C>T, p.(Arg345*)	c.979C>T, p.(Arg327*)
chr7:114292331-114292332delCA	c.1243_1244delCA, p.(Gln415Valfs*7)	c.1168_1169delCA, p.(Gln390Valfs*7)	c.1219_1220delCA, p.(Gln407Valfs*7)	c.1165_1166delCA, p.(Gln389Valfs*7)
chr7:114298286C>T	c.1507C>T, p.(Pro530Leu)	c.1432C>T, p.(Arg478*)	c.1483C>T, p.(Arg495*)	c.1429C>T, p.(Arg477*)
chr7:114299688G>C	c.1682G>C, p.(Arg561Pro)	c.1607G>C, p.(Arg536Pro)	c.1658G>C, p.(Arg553Pro)	c.1604G>C, p.(Arg535Pro)
chr7:114299695delT	c.1689delT, p.(Phe563Leufs*28)	c.1614delT, p.(Phe538Leufs*28)	c.1665delT, p.(Phe555Leufs*28)	c.1611delT, p.(Phe537Leufs*28)
chr7:114302130G>A	c.1733G>A, p.(Arg578His)	c.1658G>A, p.(Arg553His)	c.1709G>A, p.(Arg570His)	c.1655G>A, p.(Arg552His)
chr7:114302162C>T	c.1765C>T, p.(Arg589*)	c.1690C>T, p.(Arg564*)	c.1741C>T, p.(Arg581*)	c.1687C>T, p.(Arg563*)



Supplementary figure S1 FOXP2 isoforms. According to RefSeq FOXP2 has 6 isoforms. Isoform I is reported to be the major one [1] and is characterized best, isoform II is the presumably longest. Isoform schemes are displayed as shown in the Ensemble genome browser, basepair and amino acid numbers are according to RefSeq NM numbers. We annotated all published and new mutations to isoform II as this one is currently used in the ExAC browser and the DECIPHER database. To better allow comparability with previously published mutations named after isoform I, we annotated all mutations to the four longest isoforms I, II, IV and V in supplementary table S1.

1 Lai CS, Fisher SE, Hurst JA, Vargha-Khadem F, Monaco AP. A forkhead-domain gene is mutated in a severe speech and language disorder. *Nature* 2001;**413**(6855):519-23.