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Supplemental Data

Mutations in *HPSE2* Cause Urofacial Syndrome

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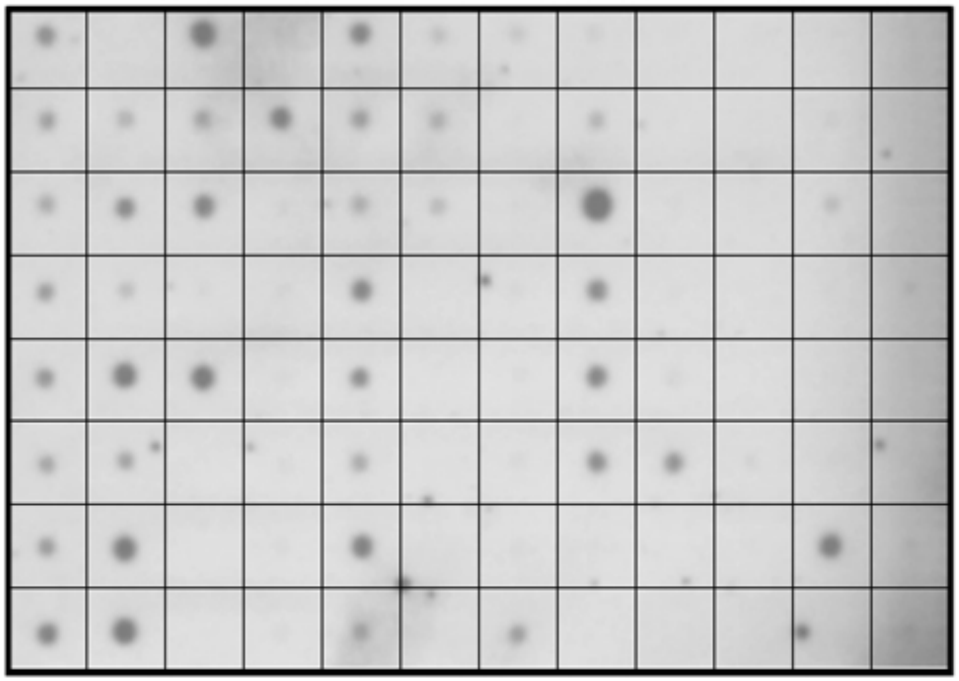

	1	2	3	4	5	6	7	8	9	10	11	12
A	whole brain		substantia nigra	heart	esophagus	colon, transverse	kidney	lung	liver	leukemia HL-60	fetal brain	yeast total RNA
B	cerebral cortex	cerebellum right	nucleus accumbens	aorta	stomach	colon, descending	skeletal muscle	placenta	pancreas	HeLa S3	fetal heart	yeast tRNA
C	frontal lobe	corpus callosum	thalamus	atrium, left	duodenum	rectum	spleen	bladder	adrenal gland	Leukemia K-562	fetal kidney	*E.Coli* rRNA
D	parietal lobe	amygdala	pituitary gland	atrium, right	jejunum		thymus	uterus	thyroid	Leukemia MOLT-4	fetal liver	*E.Coli* DNA
E	occipital lobe	caudate nucleus	spinal cord	ventricle, left	ileum		peripheral blood leukocyte	prostate	salivary gland	Burkitt's lymphoma Raji	fetal spleen	poly r(A)
F	temporal lobe	hippo-campus		ventricle, right	ileocecum		lymph node	testis	mammary gland	Burkitt's lymphoma Daudi	fetal thymus	human C_0t-1 DNA
G	pg* of cerebral cortex	medulla oblongata		inter-ventricular septum	appendix		bone marrow	ovary		colorectal adeno carcinoma SW 480	fetal lung	human DNA 100ng
H	pons	putamen		apex of the heart	colon, ascending		trachea			lung carcinoma A549		human DNA 500ng

* Paracentral gyrus

Figure S1. *HPSE2* RNA Distribution Profiling by Dot Blot Analysis
The human multiple tissue RNA dot-blot indicating expression of *HPSE2* in a number of tissues, notably adult brain, bladder, uterus, prostate and testis.

Table S1. Sequence of Primers for Amplification of Exon-Intron Boundaries and Break-Point Deletion Boundaries in *HPSE2*

Fragment	Oligonucleotide	Size (bp)
Primers for PCR and Sequence Analysis		
HPSE2 exon 1	Forward: cactagcgagacccagtagga	394
	Reverse: ggcttgaggggtgttacta	
HPSE2 exon 2	Forward: gccctcagggagtaggaaga	293
	Reverse: cttccgctccccaaataaa	
HPSE2 exon 3	Forward: ggagttggagagccttctga	399
	Reverse: cagacagatgtgtaccccaaaa	
HPSE2 exon 4	Forward: agtgggaagctcatagaaagg	378
	Reverse: ttctgggccagggtactaga	
HPSE2 exon 5	Forward: aaaggcagagagatctgtgga	400
	Reverse: gggtgaagccactatggaaa	
HPSE2 exon 6	Forward: tcagtttatttccttttgatttaggg	243
	Reverse: ttttcctggaggatgaagga	
HPSE2 exon 7	Forward: gcaacagagaccttgggcta	250
	Reverse: tgggactttgtgtctttttcc	
HPSE2 exon 8	Forward: cccctgaaaacaggaaatca	300
	Reverse: gcagaataatcagcaacacca	
HPSE2 exon 9	Forward: ttggtgactggagattggtg	295
	Reverse: tggaacaaggcatgatcaaa	
HPSE2 exon 10	Forward: agtgatttctggcccgtgt	382
	Reverse: ggtgattccagaggcaaaaa	
HPSE2 exon 11	Forward: tagggtctatggtggccaag	400
	Reverse: cctactccatcccactgagc	
HPSE2 exon 12	Forward: tgtcagctgtgtgtgtgtatcag	377
	Reverse: ggctggttgctaggatgtct	
HPSE2 exon 12b	Forward: cttcaccatgcttcctggtt	334
	Reverse: gcacagtcaaacacgagttca	
Primers for Exon8-9 Deletion Breakpoint Analysis		
HPSE2 5' exon 8	Forward: cgccctttatcaggtcacat	
HPSE2 3' exon 9	Reverse: gtccggcaaatgtaagcaat	

All primers amplified at an annealing temperature of 60°C