Gene	Location	Variant	gnomAD AF [1]	CADD [2]	Revel [3]	Associated Phenotypes	Interpretation
LOXHD1	18:44,056,924-44,237,183	c.1763 G>C; p.G579A	0	25.2	0.822	**Recessive** Nonsyndromic Hearing Loss [4]; Dominant Late-Onset Fuch's [5]	VUS
SLC16A12	10:91,190,056-91,295,351	c.113 G>A; p.R38Q	$1.6E10^{-5}$	13.71	0.105	Dominant Juvenile and Age-Related Cataracts, Microcornea, and Renal Glucosuria [6, 7]	VUS

Table S1: Genetic variants segregating in all three affected family members, their locations (hg19), their gnomAD allele frequencies, CADD and REVEL scores, and associated phenotypes. VUS is variant of uncertain significance.

Gene	Location	Human LOF Phenotype
CRYL1	13:20,977,808-21,099,996	Hearing Loss [8, 9]
IFT88	13:21,141,296-21,265,583	Retinal Degeneration; NS CL/CP [10, 11]
IL17D	13:21,275,652-21,297,237	N/A
EEF1AKMT1	13:21,302,870-21,348,100	N/A
XPO4	13:21,351,468-21,476,913	Laryngomalacia, Developmental Delay [8, 12]
LATS2	13:21,547,175-21,635,725	Cardiac Defects, Cancer Development [12, 13]
SAP18	13:21,140,119-21,149,097	N/A
SKA3	13:21,727,734-21,750,691	N/A
MRP63	13:21,750,797-21,753,223	N/A
MIPEPP3	13:21,872,264-21,967,062	N/A
ZDHHC20	13:21,946,710-22,033,442	N/A
MICU2	13:21,946,710-22,033,442	Neurodevelopmental Disorder [14]

Table S2: Genes within the microdeletion, their location (hg19), and any suggested loss of function (LOF) human phenotypes.

GO Biological Process	Fold Enrichment	Raw P-Value	FDR
External Encapsulating Structure Organization	8.73	2.93E-15	1.53E-11
Extracellular Structure Organization	8.79	2.53E-15	1.99E-11
Extracellular Matrix Organization	8.82	2.35E-15	3.69E-11
Tube Development	3.49	4.09E-09	1.60E-05
Blood Vessel Development	4.44	1.01E-08	3.18E-05
Vasculature Development	4.26	2.09E-08	4.69E-05
Collagen Fibril Organization	15.44	2.03E-08	5.30E-05
Tube Morphogenesis	3.52	2.72E-07	4.74E-04
Collagen Metabolic Process	13.5	3.23E-07	5.06E-04
Blood Vessel Morphogenesis	4.4	2.71E-07	5.31E-04
Cellular Response to Amino Acid Stimulus	11.13	1.24E-06	1.49E-03
Angiogenesis	4.72	1.21E-06	1.58E-03
Circulatory System Development	2.94	1.17E-06	1.67E-03
Ossification	4.97	1.57E-06	1.75E-03
Eye Development	4.29	1.77E-06	1.85E-03
Visual System Development	4.25	2.01E-06	1.97E-03
Cellular Response to Acid Chemical	10.04	2.52E-06	1.97E-03
Tissue Development	2.27	2.29E-06	2.00E-03
Sensory System Development	4.18	2.43E-06	2.00E-03
Animal Organ Morphogenesis	2.77	2.19E-06	2.02E-03
Response to Wounding	3.92	2.73E-06	2.04E-03
Anatomical Structure Morphogenesis	2.07	3.32E-06	2.36E-03
Cellular Response to Transforming Growth Factor Beta Stimulus	6.68	4.40E-06	3.00E-03
Odontogenesis	7.35	6.48E-06	3.77E-03
Regulation of Collagen Metabolic Process	14.71	6.45E-06	3.89E-03
Eye Morphogenesis	6.43	6.06E-06	3.96E-03
Response to Transforming Growth Factor Beta	6.39	6.38E-06	4.00E-03
Wound Healing	4.26	8.50E-06	4.76E-03
Cell Adhesion	2.66	1.10E-05	5.95E-03
Positive Regulation of Collagen Biosynthetic Process	19.06	1.28E-05	6.68E-03
Positive Regulation of Collagen Metabolic Process	18.38	1.49E-05	7.32E-03
Negative Regulation of Developmental Process	2.68	1.48E-05	7.48E-03
Supramolecular Fiber Organization	3.19	2.04E-05	9.70E-03
Enzyme-Linked Receptor Protein Signaling Pathway	3.06	2.16E-05	9.98E-03
Response to Amino Acid	7.29	2.27E-05	1.02E-02
Camera-Type Eye Development	4.01	3.35E-05	1.46E-02
Regulation of Collagen Biosynthetic Process	13.91	4.99E-05	2.12E-02
Response to Acid Chemical	6.43	5.25E-05	2.16E-02
System Development	1.64	5.42E-05	2.18E-02
Anatomical Structure Formation Involved in Morphogenesis	2.52	5.72E-05	2.24E-02
Sensory Organ Development	3.01	7.22E-05	2.76E-02
Cellular Process	1.16	7.75E-05	2.89E-02
Response to Endogenous Stimulus	2.18	8.92E-05	3.25E-02
Regulation of Biomineralization	7	9.47E-05	3.37E-02
Direct Ossification	44.12	1.00E-04	3.41E-02
Regulation of Smooth Muscle Cell Proliferation	5.8	1.05E-04	3.41E-02
Intramembranous Ossification	44.12	1.00E-04	3.49E-02
Osteoblast Differentiation	5.8	1.05E-04	3.49E-02
Multicellular Organism Development	1.56	1.46E-04	4.68E-02

Table S3: All significant Gene Ontology (GO) processes [15] for upregulated genes with an adjusted p-value of less than or equal to 1e-10. FDR is false discovery rate.

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