

Supplementary Table 1. Individuals with deletions in chromosome 4 including the *NFKB1* gene reported in Decipher database (<https://www.deciphergenomics.org/>).

Patient ID	Sex	Chr 4 deletion (coordinates, GRCh38)	Size	Inheritance / Genotype	Phenotype (s)	Age at last clinical assessment
1580	male	4:98747288-106803182	8.06 Mb	Unknown / Heterozygous	Abnormal hair morphology, autism, blepharophimosis, broad nasal tip, coarse facial features, facial asymmetry, flat occiput, infra-orbital crease, intellectual disability, sleep disturbance, uplifted earlobe	8 years
249234	female	4:100010584-104308722	4.30 Mb	De novo / Heterozygous	Coarse facial features, intellectual disability, macrotia, Scoliosis, Seizure	10 years
253148	male	4:98109249-103106128	5.00 Mb	De novo / Heterozygous	n.a.	16 years
263047	female	4:99253457-103085561	3.83 Mb	Unknown / Heterozygous	Autism, intellectual disability	16 years
269925	male	4:97840809-108269099	10.43 Mb	De novo / Heterozygous	n.a.	1 year
303816	female	4:98416654-103719753	5.30 Mb	Unknown / Heterozygous	Abnormal facial shape, decreased circulating antibody level , global developmental delay, short foot	17 years
314461	female	4:102152033-104580942	2.43 Mb	De novo / Heterozygous	Delayed gross motor development, short stature	1 year
384471	male	4:102103901-103020291	916 Kb	Unknown / Heterozygous	Global developmental delay	4 years
400942	female	4:101921665-105168081	3.25 Mb	De novo / Heterozygous	Abnormal palate morphology, abnormality of the outer ear, abnormality of toe, arachnodactyly, delayed speech and language development, fine hair, finger clinodactyly, joint laxity, long eyelashes, microdontia, narrow mouth, nasal speech, pes planus, postaxial hand polydactyly, short palpebral fissure, spotty hyperpigmentation, supernumerary tooth, wide nasal base, wide nasal bridge	4 years
402229	male	4:92221672-106721239	14.50 Mb	Imbalance arising from a balanced parental rearrangement / Heterozygous	Abnormality of the cardiovascular system, feeding difficulties in infancy, horseshoe kidney, incoordination, low posterior hairline, pectus carinatum, ptosis	19 years

Patient ID	Sex	Chr 4 deletion (coordinates, GRCh38)	Size	Inheritance / Genotype	Phenotype (s)	Age at last clinical assessment
415252	male	4:96898260-104297409	7.40 Mb	Unknown / Heterozygous	n.a.	4 years