Supplementary material for Hosono et al., "A case of childhood glaucoma with a combined partial monosomy 6p25 and partial trisomy 18p11 due to an unbalanced translocation" *Ophthalmic Genetics*, 2020.

Supplementary Table S1. Summary of clinical features in patients with 6p25 deletions.

	Our patient	Frequency of previous cases[a]
Structural eye abnormality		
Posterior embryotoxon	+	72.7% (8/11)
Iris hypoplasia	+	70.0% (7/10)
Glaucoma	+	40.0% (4/10)
Refractive error	-	84.6% (11/13)
Strabismus	+	81.3% (13/16)
Corectopia	+	75.0% (6/8)
Other anterior eye chamber anomaly[b]	+	84.2% (16/19)
Craniofacial features		
Frontal bossing/high forehead	+	100% (13/13)
Midface retrusion/flat midface	+	100% (8/8)
Downslanted palpebral fissures	−	77.8% (14/18)
Hypertelorism/telecanthus	−	95.8% (23/24)
External ear anomalies/low-set	+	82.4% (14/17)
Wide/depressed nasal bridge	+	100% (15/15)
Short philtrum	−	50.0% (4/8)
Smooth/flat philtrum	+	100% (4/4)
High/cleft palate	−[c]	66.7% (10/15)
Cognitive and physical development		
Development delay	+	100% (24/24)
Intellectual disability	+	85.7% (12/14)
Language impairment	+	94.7% (18/19)
Hypotonia	+	76.9% (10/13)
Hearing loss	+[d]	78.3% (18/23)
Heart defect	+	55.6% (10/18)

Brain abnormalities		
Dandy-Walker malformation	−	62.5% (5/8)
Hydrocephalus/Abnormal skull shape	−	84.6% (11/13)
White matter abnormalities	+	85.7% (6/7)
Musculoskeletal anomalies		
Epiphyseal dysplasia	-[e]	100% (5/5)
Hip dysplasia	-[e]	100% (3/3)
Hand anomaly	+	91.7% (11/12)
Foot anomaly	−	88.9% (16/18)
Dental abnormalities	U	69.2% (9/13)
Hernia (umbilical or inguinal)	−	30% (3/10)

[a]Linhares ND et al (17).

[b]Other anterior eye chamber anomaly: Goniodysgenesis, Axenfeld-Rieger anomaly, Axenfeld-Rieger Syndrome, coloboma, corneal opacity, cataract, iridocorneal synechiae, Duane retraction syndrome, exophthalmos, enophthalmos, iridocorneal adhesions, microphthalmia.

[c]There is no cleft palate, but the high palate is unknown.

[d]Deafness was assessed by an auditory brainstem response test.

[e]No abnormalities in orthopedic assessment.

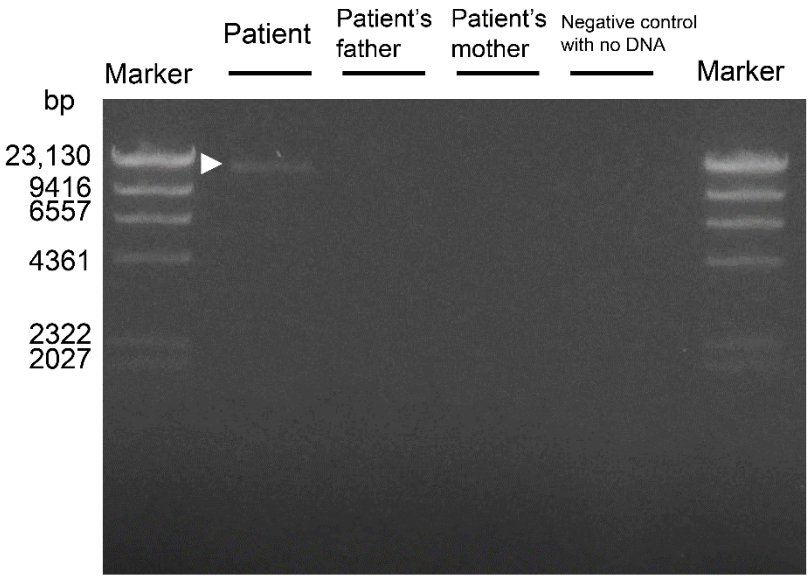
+, present; -, absent; U, unknown

Supplementary Table S2. Detailed description of genes included in the 6p25 deletion and 18p11 duplication regions.

Gene	Accession no.	Cytogenetic location	Position	Strand	Description
DUSP22	NM_001286555.1	6p25.3	chr6:292057-351355	+	Dual specificity phosphatase 22
IRF4	NM_002460.3	6p25.3	chr6:391739-411443	+	Interferon regulatory factor 4
EXOC2	NM_018303.5	6p25.3	chr6:485138-693141	-	Exocyst complex component 2
HUS1B	NM_148959.3	6p25.3	chr6:655939-656964	-	HUS1 checkpoint clamp component B
FOXQ1	NM_033260.3	6p25.3	chr6:1312675-1314993	+	Forkhead box Q1
FOXF2	NM_001452.1	6p25.3	chr6:1390069-1395832	+	Forkhead box F2
FOXC1	NM_001453.2	6p25.3	chr6:1610681-1614132	+	Forkhead box C1
GMDS	NM_001500.3	6p25.3	chr6:1624035-2245868	-	GDP-mannose 4,6-dehydratase
MYLK4	NM_001012418.4	6p25.2	chr6:2663863-2751200	-	Myosin light chain kinase family member 4
WRNIP1	NM_020135.2	6p25.2	chr6:2765666-2785979	+	Werner helicase interacting protein 1
SERPINB1	NM_030666.3	6p25.2	chr6:2832566-2842283	-	Serpin family B member 1
SERPINB9	NM_004155.5	6p25.2	chr6:2887499-2903546	-	Serpin family B member 9
SERPINB6	NM_004568.5	6p25.2	chr6:2948393-2972399	-	Serpin family B member 6
NQO2	NM_000904.4	6p25.2	chr6:3000050-3020110	+	NAD(P)H quinone dehydrogenase 2
RIPK1	NM_003804.4	6p25.2	chr6:3064122-3115421	+	Receptor interacting serine/threonine kinase 1
BPHL	NM_004332.3	6p25.2	chr6:3118610-3153432	+	Biphenyl hydrolase like
TUBB2A	NM_001069.2	6p25.2	chr6:3153900-3157783	-	Tubulin beta 2A class IIa
TUBB2B	NM_178012.4	6p25.2	chr6:3224495-3227968	-	Tubulin beta 2A class IIb
PSMG4	NM_001128591.1	6p25.2	chr6:3259162-3268300	+	Proteasome assembly chaperone 4

SLC22A23	NM_015482.1	6p25.2	chr6:3269207-3456793	-	Solute carrier family 22 member 23
PXDC1	NM_183373.3	6p25.2	chr6:3722836-3752246	-	PX domain containing 1
FAM50B	NM_012135.2	6p25.2	chr6:3849600-3851554	+	Family with sequence similarity 50 member B
PRPF4B	NM_003913.4	6p25.2	chr6:4021569-4065217	+	Pre-mRNA processing factor 4B
FAM217A	NM_173563.2	6p25.2	chr6:4068593-4079457	-	Family with sequence similarity 217 member A
C6orf201	NM_001085401.2	6p25.2	chr6:4079440-4130999	+	Chromosome 6 open reading frame 201
ECI2	NM_206836.2	6p25.2	chr6:4115927-4135831	-	Enoyl-CoA delta isomerase 2
CDYL	NM_004824.3	6p25.1	chr6:4776680-4955778	+	Chromodomain Y-like
USP14	NM_005151.3	18p11.32	chr18:158483-213739	+	Ubiquitin specific peptidase 14
THOC1	NM_005131.2	18p11.32	chr18:214520-268059	-	THO complex 1
COLEC12	NM_130386.2	18p11.32	chr18:319355-500729	-	Collectin subfamily member 12
CETN1	NM_004066.2	18p11.32	chr18:580343-582020	+	Centrin 1
CLUL1	NM_014410.4	18p11.32	chr18:596998-650298	+	Clusterin like 1
TYMSOS	NM_001012716.2	18p11.32	chr18:649620-658340	-	TYMS opposite strand
TYMS	NM_001071.2	18p11.32	chr18:657604-673499	+	Thymidylate synthetase
ENOSF1	NM_202758.3	18p11.32	chr18:670324-712517	-	Enolase superfamily member 1
YES1	NM_005433.3	18p11.32	chr18:721592-812327	-	YES proto-oncogene 1, Src family tyrosine kinase
ADCYAP1	NM_001099733.1	18p11.32	chr18:904944-912173	+	Adenylate cyclase activating polypeptide 1
METTL4	NM_022840.4	18p11.32	chr18:2537524-2571502	-	Methyltransferase like 4
NDC80	NM_006101.3	18p11.32	chr18:2571556-2616634	+	NDC80 kinetochore complex component
SMCHD1	NM_015295.2	18p11.32	chr18:2655886-2805015	+	Structural maintenance of chromosomes flexible hinge domain containing 1

EMILIN2	NM_032048.2	18p11.32 - 18p11.31	chr18:2847004-2916001	+	Elastin microfibril interfacer 2
LPIN2	NM_014646.2	18p11.31	chr18:2916992-3011945	-	Lipin 2
MYOM1	NM_003803.3	18p11.31	chr18:3066805-3220106	-	Myomesin 1
MYL12A	NM_006471.3	18p11.31	chr18:3247528-3256235	+	Myosin light chain 12A
MYL12B	NM_001144945.1	18p11.31	chr18:3262717-3278282	+	Myosin light chain 12B
TGIF1	NM_173208.2	18p11.31	chr18:3449649-3458409	+	TGFB induced factor homeobox 1
DLGAP1	NM_004746.3	18p11.31	chr18:3496030-4455266	-	DLG associated protein 1
AKAIN1	NM_001145194.1	18p11.31	chr18:5143672-5197255	-	A-kinase anchor inhibitor 1
ZBTB14	NM_001143823.2	18p11.31	chr18:5289018-5296194	-	Zinc finger and BTB domain containing 14
EPB41L3	NM_012307.3	18p11.31	chr18:5392380-5544309	-	Erythrocyte membrane protein band 4.1 like 3
TMEM200C	NM_001080209.1	18p11.31	chr18:5890184-5892103	-	Transmembrane protein 200C
L3MBTL4	NM_173464.3	18p11.31	chr18:5954705-6414910	-	L3MBTL histone methyl-lysine binding protein 4
ARHGAP28	NM_001010000.2	18p11.31	chr18:6834432-6915715	+	Rho GTPase activating protein 28
LAMA1	NM_005559.3	18p11.31	chr18:6941743-7117813	-	Laminin subunit alpha 1
LRRC30	NM_001105581.1	18p11.23	chr18:7231137-7232042	+	Leucine rich repeat containing 30
PTPRM	NM_001105244.1	18p11.23	chr18:7567314-8406859	+	Protein tyrosine phosphatase, receptor type M
RAB12	NM_001025300.2	18p11.22	chr18:8609443-8639380	+	RAB12, member RAS oncogene family
MTCL1	NM_015210.3	18p11.22	chr18:8717369-8832775	+	Microtubule crosslinking factor 1
NDUFV2	NM_021074.4	18p11.22	chr18:9102628-9134343	+	NADH:ubiquinone oxidoreductase core subunit V2



Supplementary Figure S1. Results of long-range PCR analysis.

Long-range PCR was performed to amplify junction fragments containing the translocation breakpoints. We successfully amplified approximately 20-kbp of the product (indicated by arrow) using the specially designed primer pair (see text). The amplification conditions were as follows: PCR was performed using the Tks Gflex DNA Polymerase kit (Takara, Shiga, Japan) for 30 cycles at 98°C for 10s, 60°C for 30s, and 68°C for 10 min in an automated thermal cycler.