Supplementary Material

Genetic Imbalance in Patients with Cervical Artery Dissection

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1. CADISP (Cervical Artery Dissections and Ischemic Stroke Patients) Co-investigators:

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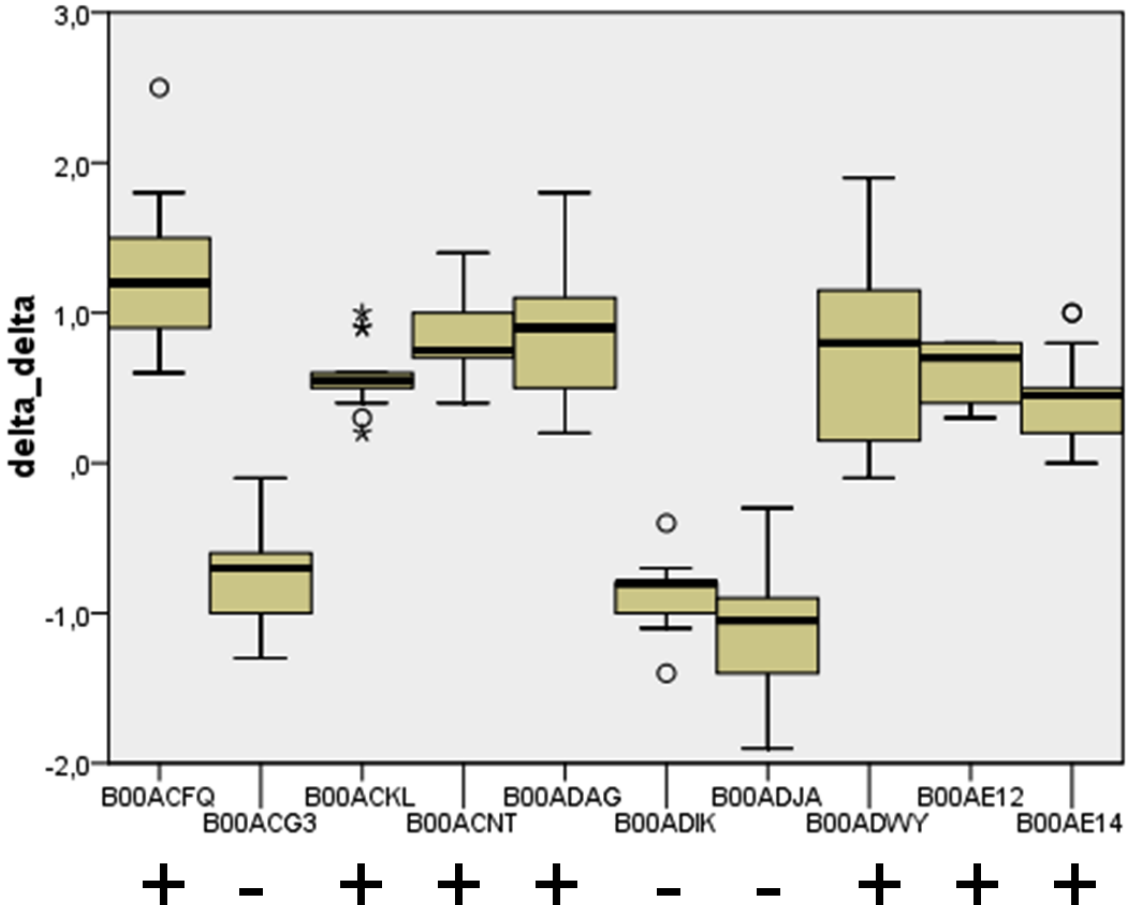
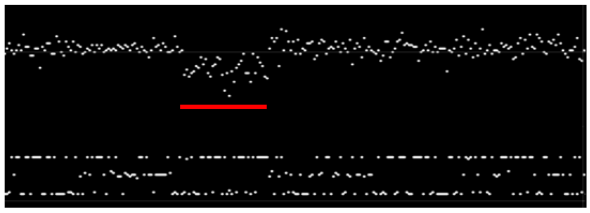
2. Validation of 10 randomly selected CNV findings by Sybr Green quantitative PCR. Box plots visualize delta-delta values for patient and three control individuals (six replicas). + indicates putative duplication (gain), - indicates putative deletion (loss).



3. Validation of 5 randomly selected CNV findings by PCR-amplification and sequencing analysis of breakpoint-joining fragments.

1. Visual inspection of putative deletion in patient B00ADZH with noise-free-CNV software [16].
(start-SNP: 8:13,968,625; end-SNP: 8:14,086,273 – positions according to GRCh37.p13)

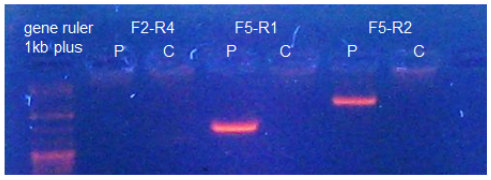


LogR Ratio (signal intensities)

Bar (▬) indicates deletion harboring SNPs with reduced signal intensities and loss of heterozygosity

Minor allele frequencies

2. Identify breakpoints-joining region by PCR amplification.



Six different sequences upstream of the first SNP within the putative deletion were used as forward primers. Six sequences downstream of the end SNP were used as reversed primer. 36 combinations of primers were tested by PCR of DNA from patients (P) and referents (C). DNA fragments between primers F5 and R1 or R2 appear to cover the deletion breakpoint.

3. Compare sequence of joining fragment with reference sequence to identify deletion breakpoints at positions 8:13,966,251 and 8:14,087,785.

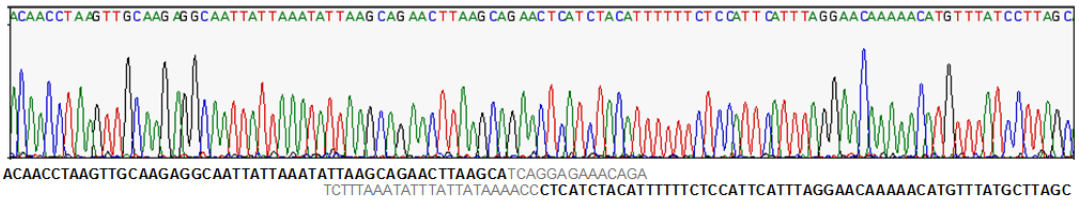


Table. Breakpoint identification and validation of five putative PennCNV-findings. Positions of start-SNP and end-SNPs of putative CNV calls as well as position of identified breakpoints in sequence of joining fragment were mapped on the GRCh17.p13 version of the human genome, in accordance with the SNP-annotation of the used Illumina platform.

			PennCNV	PennCNV	Estimated	Confirmed	Confirmed	True
Patient	**CN-state**	**Chr.**	**Start-Position**	**End-position**	**Length**	**Breakpoint**	**Breakpoint**	**Length**
B00ADWN	1	3	111.897.540	112.056.934	159.394	111.886.897	112.055.840	168.943
B00ADWN	1	9	108.395.195	108.474.679	79.484	108.390.568	108.478.065	87.497
B00ADXF	1	9	703.725	928.456	224.731	696.335	926.339	230.004
B00ADZD	3	19	1.989.867	2.677.012	687.145	2.021.905	2.681.089	659.184
B00ADZH	1	8	13.968.625	14.086.273	117.648	13.966.251	14.087.785	121.534