Diagnosis of Dyskeratosis Congenita and Aplastic Anemia – *PRDM8*

Dyskeratosis Congenita

Dyskeratosis congenita (DKC) is associated with impaired telomere maintenance and with clinical features of premature aging. So far, diagnosis of DKC relies particularly on telomere length measurements, but not all patients reveal significant telomere attrition raising the need for additional biomarkers.

Aplastic Anemia

Acquired aplastic anemia (AA) is another bone marrow failure syndrome that can mimic clinical features of DKC and is likewise characterized by telomere attrition.

How does it work?

Dyskeratosis congenita and aplastic anemia have aberrant DNA-methylation at an internal promoter region of PR domain containing 8 (*PRDM8*). We provide service for analysis of DNA-methylation at the relevant genomic region by MassARRAY.

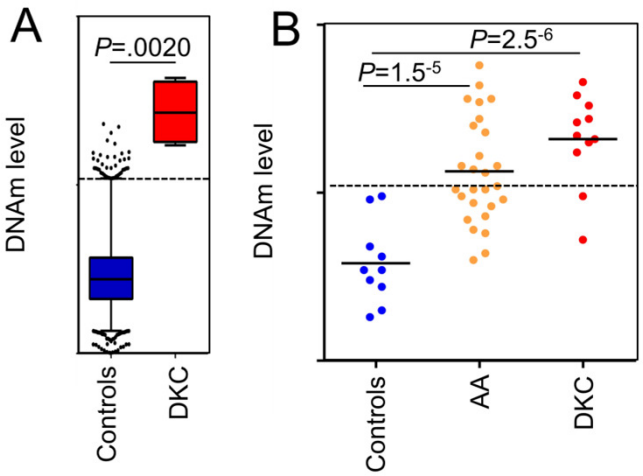

Figure 1: DNA-methylation in PRDM8. *(A) Distribution of DNA-methylation levels in 4,131 control samples of normal peripheral blood (99 percentile is indicated by dotted line) and in four DKC samples. (B) MassARRAY analysis validated significantly higher DNA-methylation levels in blood of patients with dyskeratosis congenita (n=27) and aplastic anemia (n=11) as compared to normal controls (n=10) .*

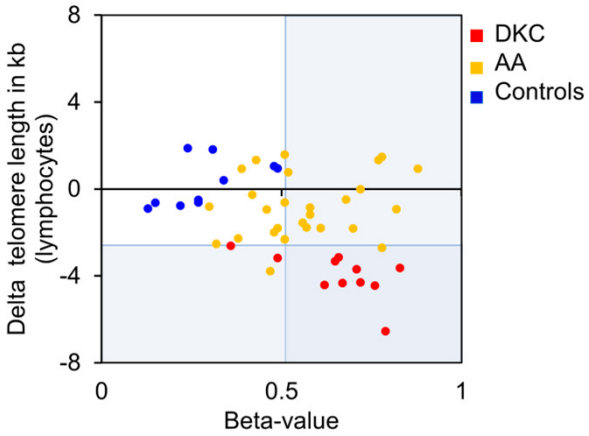

Figure 2: DNA-methylation in PRDM8 and telomere length are complementary. *Telomere length of lymphocytes was measured by flow-FISH. Delta telomere length – in relation to mean telomere length of healthy controls – was plotted against DNA-methylation in PRDM8 (blue lines resemble 99 percentiles). Importantly, telomere length does not correlate with DNA-methylation in PRDM8. Therefore the two methods are complementary to also identify patients with normal telomere length.*

Our service for you:

- You send us blood samples (EDTA or Heparin) or genomic DNA. Shipment can be performed at room temperature (if possible >1ml blood or >200 ng DNA)
- We perform bisulfite conversion of DNA
- We analyze the DNA-methylation by MassARRAY
- We determine the DNA methylation level at the relevant genomic region
- Results – including raw data, and graphical presentation – are provided by Email (usually within two weeks)

Publications:

- Weidner CI et al. ***Oncotarget.*** 2016 doi:10.18632/oncotarget
- Patent application: DE 10 2015 121 969.7

Further Information

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