Cat Nr/REF: KBI-40101

English

For professional use only

Poseidon™ Repeat Free™ Miller-Dieker LIS (17p13) & Smith-Magenis RAI (17p11) probe

Introduction:

The **Miller-Dieker** lissencephaly syndrome appears to be caused by deletion of several genes on 17p. Deletion of or mutation in the LIS1 gene appears to cause the lissencephaly. On the other hand, facial dysmorphism and other anomalies in Miller-Dieker patients appear to be the consequence of deletion of additional genes distal to LIS1. About 15% of patients with isolated lissencephaly and more than 90% of patients with Miller-Dieker syndrome have microdeletions in a critical 350-kb region at 17p13.3.

Smith-Magenis Syndrome is a distinct and clinically recognizable contiguous gene syndrome characterized by a specific pattern of physical, behavioral and developmental features. It is caused by a deletion of chromosome 17, referred to as deletion 17p11.2. The RAI1 gene region has been identified to be deleted in more than 90% of Smith-Magenis syndrome patients.

Intended use:

The **Miller-Dieker** region probe is optimized to detect copy numbers of the PAFAH1B1 gene (LIS1) region at 17p13.3. The Smith-Magenis RAI1 region probe at 17p11.2 is serving as internal control.
The **Smith-Magenis** region probe is optimized to detect copy numbers of the RAI1 gene region described to be involved in Smith-Magenis syndrome at 17p11.2. The Miller-Dieker LIS1 probe at 17p13.3 is serving as internal control

The probe is recommended to be used in combination with a Poseidon FISH Kit providing necessary reagents to perform FISH (KBI-60002, KBI-60003 or KBI-60001) for optimal results.

Critical region 1 (red):

The **Miller-Dieker** specific DNA probe is direct-labeled with Platinum*Bright*550.

Critical region 2 (green):

The **Smith-Magenis** specific DNA probe is direct-labeled with Platinum*Bright*495.

Reagent:

Poseidon probes are direct-labeled DNA probes provided in a ready-to-use format. Apply 10 µl of probe to a sample area of approximately 22 x 22 mm.

Please refer to the Instructions for Use for the entire Poseidon FISH protocol.

Poseidon Repeat Free probes do not contain Cot-1 DNA. Hybridization efficiency is therefore increased and background, due to unspecific binding, is highly reduced.

Interpretation:

The Miller-Dieker and Smith-Magenis probe is designed as a dual-color assay to detect deletions at 17p13 and 17p11 respectively. Deletions at the **Miller-Dieker** region will show one red signal and two signals at the Smith-Magenis control region (1R2G). Deletions at the **Smith-Magenis** region will show one green and two red signals at the Miller-Dieker control region (2R1G). Two single color red (R) and green (G) signals will identify the normal chromosomes 17 (2R2G)

	Normal Signal Pattern	Del 17(17p13.3) MDCR	Del 17(17p11.2) SMCR
Expected Signals	2R2G	1R2G	2R1G

References:

Miller-Dieker / Lissencephaly:
Miller, J, 1963, Neurology 13; 841-850
Dieker H, 1969, Birth Defects, 5(2); 53-64
Vlangos et al, 2005 Am.J.Med.Genet. 132A; 278-282

Smith-Magenis:
Smith A, 1986, Am.J.Med.Genet. 24 ; 393-414
Cardoso et al, 2002, Hum Mutat. Jan; 19(1); 4-15

POSEIDON™
REPEAT-FREE™ FISH PROBES

Application Manual

KBI-40101
MD Miller-Dieker LIS (17p13) / Smith-Magenis RAI (17p11)

IVD
for EU only

CE

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2°C 8°C long term storage

KREATECH DIAGNOSTICS

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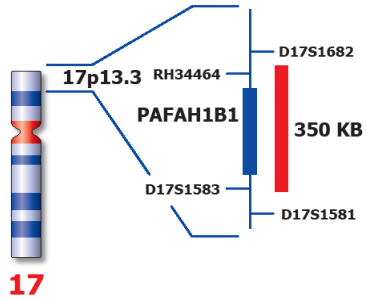

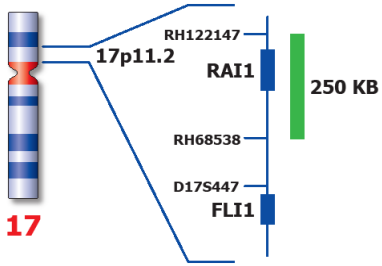

Not to scale

Application manual