

Kreatech™ FISH probes Product Information Sheet

KBI-40101

Miller-Dieker PAFAH1B1 (17p13) /

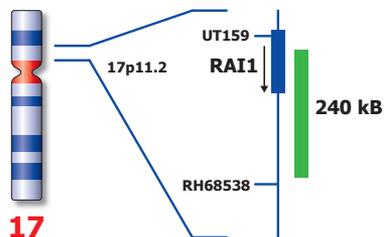
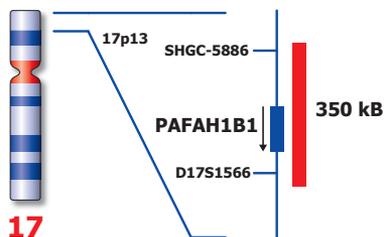
Smith-Magenis RAI1 (17p11)



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Not to scale

Kreatech™ Miller-Dieker PFAH1B1 (17p13) / Smith-Magenis RAI1 (17p11) FISH probe

- Introduction:** The **Miller-Dieker** lissencephaly syndrome appears to be caused by deletion of several genes on 17p. Deletion of or mutation in the PFAH1B1 (previously known as 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 PFAH1B1. 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 (previously known as SMCR, KIAA1820 or SMS) gene region has been identified to be deleted in more than 90% of Smith-Magenis syndrome patients.
- Intended use:** The **Miller-Dieker PFAH1B1** region probe is optimized to detect copy numbers of the PFAH1B1 region at 17p13. The **Smith-Magenis RAI1** region probe is optimized to detect copy numbers of the RAI1 gene region at 17p11.
- The probe is recommended to be used in combination with one of the Kreatech Pretreatment kits providing necessary reagents to perform FISH on various sample types for optimal results. (see also www.LeicaBiosystems.com and look for Kits & reagents)
- Critical region 1 (red):** The **Miller-Dieker PFAH1B1** specific FISH probe is direct-labeled with PlatinumBright™550.
Critical region 2 (green): The **Smith-Magenis RAI1** specific FISH probe is direct-labeled with PlatinumBright™495.
- Reagent:** Kreatech 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 Kreatech FISH protocol.**
- Kreatech FISH probes are REPEAT-FREE™ and therefore do not contain Cot-1 DNA. Hybridization efficiency is increased and background, due to unspecific binding, is highly reduced.**
- Interpretation:** The **Miller-Dieker PFAH1B1** and **Smith-Magenis RAI1** FISH probe are designed as a dual-color assay to detect deletions at 17p13 and 17p11 respectively. Deletions at the PFAH1B1 region will show one red signal and two signals at the RAI1 region (1R2G). Deletions at the RAI1 region will show one green and two red signals at the PFAH1B1 region (2R1G). Two single color red and green signals will identify the normal chromosomes 17 (2R2G)
- | | Normal Signal Pattern | Del(17p13) PFAH1B1 | Del(17p11) RAI1 |
|------------------|-----------------------|--------------------|-----------------|
| Expected Signals | 2R2G | 1R2G | 2R1G |
- References:** Miller, J, 1963, Neurology 13; 841-850
 Dieker H, 1969, Birth Defects, 5(2); 53-64, 278-282
 Cardoso et al, 2002, Hum Mutat. Jan; 19(1); 4-15
 Smith A, 1986, Am.J.Med.Genet. 24 ; 393-414
- Warning and precautions:** In case of emergencies check SDS sheets for medical advice. SDS sheets may be obtained by either contacting Leica Technical Support or visiting www.LeicaBiosystems.com. DNA probes contain formaldehyde which is a teratogen; do not inhale or allow skin contact. Wear gloves and a lab coat when handling DNA probes. All materials should be disposed of according to your institution's guidelines for hospital waste disposal.
- Reagent Storage and Handling:** Store at 2-8 °C. Reagents should not be used after the expiration date on the vial label.
- TECHNICAL SUPPORT** Technical support is available at www.LeicaBiosystems.com or +31 20 6919181 or via e-mail: kreatech-support@leicabiosystems.com.
- CUSTOMER SERVICE** Kreatech probes may be ordered through Leica Customer Service +31 20 6919181 or order via e-mail: purchase.orders@leica-microsystems.com.