

English

Poseidon™ STS (Xp22) & KAL (Xp22) & SE X Control probe

Introduction: X-linked ichthyosis is the result of steroid sulfatase (STS) deficiency. The majority of STS-deficient patients have a complete deletion of the STS gene, while a minority of cases can be dedicated to certain point mutations in the STS gene.

Deletion of the human KAL gene is responsible for the X chromosome-linked Kallmann's syndrome, which consists of an association between hypogonadotropic hypogonadism and anosmia (or hyposmia).

Intended use: The **STS (Xp22)** region probe is optimized to detect copy numbers of the STS gene region at Xp22. The **KAL (Xp22)** region probe is optimized to detect copy numbers of the KAL gene region at Xp22. The Chromosome X Satellite Enumeration (SE) probe is included to facilitate chromosome identification.

The probe is recommended to be used in combination with one of the Poseidon Pretreatment kits providing necessary reagents to perform FISH on various sample types for optimal results. (see also www.kreatech.com and look for Kits & reagents)

Critical region 1 (red): The **KAL (Xp22)** specific DNA probe is direct-labeled with PlatinumBright[®]550.

Critical region 2 (green): The **STS (Xp22)** specific DNA probe is direct-labeled with PlatinumBright[®]495.

Control region 3 (blue): The **SE X** control probe is direct-labeled with PlatinumBright[®]415.

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.

Interpretation: The **STS (Xp22)** and **KAL (Xp22)** probe is designed as a triple-color assay to detect deletions at two different regions at Xp22. Deletions involving the KAL region will show one red signal, two green, and two blue signals STS and SE X control region (1R2G2B). Deletions involving the STS region will show one green signal, and two red and two blue signals KAL and SE X control region (1R2G2B). Deletions involving both the KAL and STS region will show one green and one red signal and two blue signals SE X control region (1R2G2B). Two single color red (R) green (G), and blue (B) signals will identify the normal chromosomes X (2R2G2B) in females or (1R1G1B) in males.

Note*: Both genes, STS and KAL, have homologous non-functional pseudogene sequences at Yp (see Meroni et al, 1996). Very weak cross-hybridization of the Xp22 probes with Yp could be visible. It is recommended to use Metaphase chromosomes for this analysis.

	Normal Signal Pattern	Del KAL (Xp22)	Del STS (Xp22)	Del STS and KAL
Expected Signals In females	2R2G2B	1R2G2B	2R1G2B	1R1G2B
Expected Signals In males*	1R1G1B	1G1B	1R1B	1B

References: Alperin et al, 1997, J. Biol. Chem 272: 20756-20763.
Meroni et al, 1996, Hum. Mol. Genet. 5: 423-431

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Application Manual

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MD STS (Xp22) / KAL (Xp22) / SE X TC



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