

## Supplementary Online Content

Spoerri I, Brena M, De Mesmaeker J, et al. The phenotypic and genotypic spectra of ichthyosis with confetti plus novel genetic variation in the 3' end of *KRT10*: from disease to a syndrome. *JAMA Dermatol*. Published online September 11, 2014. doi:10.1001/jamadermatol.2014.2526.

**eFigure 1.** Clinical Appearance of Patients With IWC

**eFigure 2.** Overview of the IWC Disease Locus

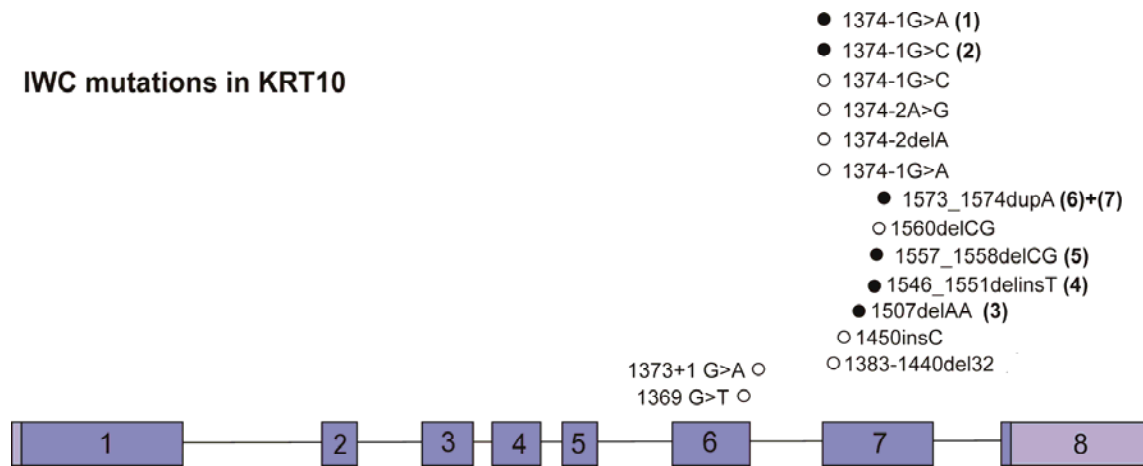
**eFigure 3.** IWC Consensus Sequence at the C-Terminus of *KRT10*

This supplementary material has been provided by the authors to give readers additional information about their work.



**Figure 1: Clinical appearance of patients with IWC.** A: Hypertrichosis on the lower arm limited to ichthyotic skin. B-C: Additional examples of typical malformation of ears. D-E: Decreased finger length relative to palms and joint contractions. F: Unguis inflexus. G: Hypoplasia of mamillae and hyperpigmentation in spots of healthy skin.

## IWC mutations in KRT10



**Figure 2: Overview of the IWC disease locus.** Schematic representation of *KRT10* exons (boxes) and introns (lines). Previously described pathological IWC mutations are indicated by empty circles [10,12]. Pathological IWC mutations, described in this study, are indicated by filled circles.

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Wt      SSGGGGRGGGSFGGGYGGSSSGGSSSGGGHGGGHGGSSSGGGYGGSSSG
P 1     R---RRTRRRKFRRRLRRRKLRRRKLRRRSRRRPRRQFRRRLRRRKLRR
P 2     R---RRTRRRKFRRRLRRRKLRRRKLRRRKLRRRSRRRPRRQFRRRLRRRKLRR
P 3     SSGGGGRGGGSFGGGYGGSSSGGSSSGGGYGGGHGGSSSGGGYGGG-LRR
P 4     SSGGGGRGGGSFGGGYGGSSSGGSSSGGGHGGGHGGSSSGGGYGGSSSG
P 5     SSGGGGRGGGSFGGGYGGSSSGGSSSGGGHGGGHGGSSSGGGYGGSSSG
P 6+7   SSGGGGRGGGSFGGGYGGSSSGGSSSGGGHGG----SSGGYGGSSSG

Wt      GSSGGGYGGSSSSGGHGGSSSGGYGGSSSGGGGGGYGGSSSGGSSSSG
P 1     RKLRRRLRGRKLQRRPRRQFQRRLRWWQFRRRRRRLRGRQLRRRQQLRR
P 2     RKLRRRLRGRKLQRRPRRQFQRRLRWWQFRRRRRRLRGRQLRRRQQLRR
P 3     RKLRRRLRGRKLQRRPRRQFQRRLRWWQFRRRRRRLRGRQLRRRQQLRR
P 4     GSSGGGYGGF--QRRPRRQFQRRLRWWQFRRRRRRLRGRQLRRRQQLRR
P 5     GSSGGGYGGSSR-RPRRQFQRRLRWWQFRRRRRRLRGRQLRRRQQLRR
P 6+7   GSSGGGYGGSSSSGGHGGKFORRRLRWWQFRRRRRRLRGRQLRRRQQLRR

Wt      GYGGGS-----SSGGHKSSSSGSGVGESSSKGPRYX
P 1     RIRRRQ-----LQRRPQVLLFRVRGRVFIX
P 2     RIRRRQ-----LQRRPQVLLFRVRGRVFIX
P 3     RIRRRQQLRRRIIRRRQQLRRRIIRRRQLQRRPQVLLFRVRGRVFIX
P 4     RIRRRQ-----LQRRPQVLLFRVRGRVFIX
P 5     RIRRRQ-----LQRRPQVLLFRVRGRVFIX
P 6+7   RIRRRQ-----LQRRPQVLLFRVRGRVFIX

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**eFigure 3: IWC consensus sequence at the C-terminus of *KRT10*.** Alignment of the wild-type (green) and mutant (black) protein sequences found in IWC patients described in this study. The first altered amino acid is marked in red. Individual polymorphisms, such as SNPs, in-frame deletions or duplications, are marked in blue. The increasing intensity of background color reflects the level of consensus between sequences.