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Positional cloning in Xp22 : towards the isolation of the gene involved in X-linked retinoschisis

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Issue Date: 1998-01-07

**Positional cloning in Xp22:
towards the isolation of the gene involved in
X-linked retinoschisis**

**Positionele klonering in Xp22:
richting de isolatie van het gen betrokken bij
X-gebonden retinoschisis**

(met een samenvatting in het Nederlands)



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**Positional cloning in Xp22:
Towards the isolation of the gene involved in
X-linked retinoschisis**

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aan de Rijksuniversiteit te Leiden,
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Voor mijn vader

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- 2.1 An Xp22.1-p22.2 YAC contig encompassing the disease loci for RS, KFSD, CLS, HYP and RP15; refined localization of RS and KFSD.
(Extended from: *Eur.J.Hum.Genet.* 4:101-104, 1996). 63
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- A CA-repeat polymorphism near DDXS418 (P122).
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Chapter 3 Cosmid-based exon trapping

- Scanning for genes in large genomic regions: cosmid-based exon trapping of multiple exons in a single product.
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Chapter 4 Isolation of retinoschisis candidate genes.

- 4.1 Characterization of a new developmental gene, *SCML1*, in Xp22.
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- 4.2 A novel human serine-threonine phosphatase related to the *Drosophila retinal degeneration C* (*rdgC*) gene is selectively expressed in sensory neurons of neural crest origin.
Hum.Mol.Genet. 6:1137-1145, 1997. 121

Chapter 5 Testing retinoschisis candidate genes

- 5.1 Exclusion of *PPEF* as the gene causing X-linked juvenile retinoschisis.
Hum.Genet. (in press) 1997. 133
- 5.2 Exclusion of the *Txp3* gene as the gene causing X-linked juvenile retinoschisis.
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