

## **Publicaties Mw. Dr. L.I. van den Born**

### [Autosomal recessive Stickler syndrome in two families caused by mutations in the COL9A1 gene.](#)

Nikopoulos K, Schrauwen I, Simon M, Collin RC, Veckeneer M, Keymolen K, Van Camp G, Cremers FP, van den Born LI.

Invest Ophthalmol Vis Sci. 2011 Mar 18. [Epub ahead of print]

### [High-Resolution Homozygosity Mapping Is a Powerful Tool to Detect Novel Mutations Causative of Autosomal Recessive RP in the Dutch Population.](#)

Collin RW, van den Born LI, Klevering BJ, de Castro-Miró M, Littink KW, Arimadyo K, Azam M, Yazar V, Zonneveld MN, Paun CC, Siemiatkowska AM, Strom TM, Hehir-Kwa JY, Kroes HY, de Faber JT, van Schooneveld MJ, Heckenlively JR, Hoyng CB, den Hollander AI, Cremers FP.

Invest Ophthalmol Vis Sci. 2011 Apr 6;52(5):2227-39. Print 2011 Apr.

### [The conclusions of Clemson et al concerning valproic acid are premature.](#)

van Schooneveld MJ, van den Born LI, van Genderen M, Bollemeijer JG.

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### [IQCB1 mutations in patients with leber congenital amaurosis.](#)

Estrada-Cuzcano A, Koenekoop RK, Coppieters F, Kohl S, Lopez I, Collin RW, De Baere EB, Roeleveld D, Marek J, Bernd A, Rohrschneider K, van den Born LI, Meire F, Maumenee IH, Jacobson SG, Hoyng CB, Zrenner E, Cremers FP, den Hollander AI.

Invest Ophthalmol Vis Sci. 2011 Feb 11;52(2):834-9. Print 2011 Feb.

### [Mutations in IMPG2, encoding interphotoreceptor matrix proteoglycan 2, cause autosomal-recessive retinitis pigmentosa.](#)

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### [Progressive loss of cones in achromatopsia: an imaging study using spectral-domain optical coherence tomography.](#)

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### [Homozygosity mapping in patients with cone-rod dystrophy: novel mutations and clinical characterizations.](#)

Littink KW, Koenekoop RK, van den Born LI, Collin RW, Moruz L, Veltman JA, Roosing S, Zonneveld MN, Omar A, Darvish M, Lopez I, Kroes HY, van Genderen MM, Hoyng CB, Rohrschneider K, van Schooneveld MJ, Cremers FP, den Hollander AI.

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[Mutations in the EYS gene account for approximately 5% of autosomal recessive retinitis pigmentosa and cause a fairly homogeneous phenotype.](#)

Littink KW, van den Born LI, Koenekoop RK, Collin RW, Zonneveld MN, Blokland EA, Khan H, Theelen T, Hoyng CB, Cremers FP, den Hollander AI, Klevering BJ.

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[A novel nonsense mutation in CEP290 induces exon skipping and leads to a relatively mild retinal phenotype.](#)

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[Comprehensive analysis of the achromatopsia genes CNGA3 and CNGB3 in progressive cone dystrophy.](#)

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[Homozygosity mapping reveals PDE6C mutations in patients with early-onset cone photoreceptor disorders.](#)

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[Identification of a 2 Mb human ortholog of Drosophila eyes shut/spacemaker that is mutated in patients with retinitis pigmentosa.](#)

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