



Transcriptional rewiring, post-transcriptional regulation and neo-functionalization: how the master sex-determining gene of medaka was born

Amaury Herpin, Cornelia Schmidt, Susanne Kneitz, Clara Gobe, Martina Regensburger, Aurélie Le Cam, Jérôme Montfort, Mateus C. Adolfi, Christina Lillesaar, Michael Kraeussling, et al.

► To cite this version:

Amaury Herpin, Cornelia Schmidt, Susanne Kneitz, Clara Gobe, Martina Regensburger, et al.. Transcriptional rewiring, post-transcriptional regulation and neo-functionalization: how the master sex-determining gene of medaka was born. Workshop: Paradigm shift in sex chromosome evolution, Leibniz Institute of Freshwater Ecology and Inland Fisheries (IGB). DEU., Sep 2019, Berlin, Germany. 42 p., 10.4126/FRL01-006417884 . hal-02371345

HAL Id: hal-02371345

<https://hal.science/hal-02371345>

Submitted on 2 Jun 2020

HAL is a multi-disciplinary open access archive for the deposit and dissemination of scientific research documents, whether they are published or not. The documents may come from teaching and research institutions in France or abroad, or from public or private research centers.

L'archive ouverte pluridisciplinaire **HAL**, est destinée au dépôt et à la diffusion de documents scientifiques de niveau recherche, publiés ou non, émanant des établissements d'enseignement et de recherche français ou étrangers, des laboratoires publics ou privés.

Amaury Herpin^{1,2}, Cornelia Schmidt², Susanne Kneitz², Clara Gobé³, Martina Regensburger², Aurélie Le Cam¹, Jérôme Montfort¹, Mateus C. Adolfi², Christina Lillesaar², Dagmar Wilhelm⁴, Michael Kraeussling², Brigitte Mourot¹, Béatrice Porcon¹, Maëlle Pannetier³, Eric Pailhoux³, Laurence Ettwiller⁵, Dirk Dolle⁵, Yann Guiguen¹ and Manfred Schartl^{2, 6, 7}

¹INRA, UR1037 Fish Physiology and Genomics, F-35000 Rennes, France

Corr. author's email: amaury.herpin@inra.fr

²University of Wuerzburg, Physiological Chemistry, Biocenter, 97074 Wuerzburg, Germany;

³UMR BDR, INRA, ENVA, Université Paris Saclay, 78350, Jouy en Josas, France;

⁴University of Melbourne, Department of Anatomy & Neuroscience, Parkville, Victoria 3010, Australia;

⁵University of Heidelberg, Centre for Organismal Studies (COS), Department of developmental Biology, Heidelberg, Germany;

⁶Comprehensive Cancer Center Mainfranken, University Hospital, 97080 Wuerzburg, Germany;

⁷Hagler Institute for Advanced Study and Department of Biology, Texas A&M University, College Station, Texas 77843, USA.

Transcriptional rewiring, post-transcriptional regulation and neo-functionalization: how the master sex-determining gene of medaka was born

The medaka *Oryzias latipes* has genetic sex determination of the XX/XY type. Dmrt1bY, the male sex-determining gene, arose through a segmental gene duplication event from an autosomal gene, dmrt1a. Both genes are co-orthologs of the highly conserved dmrt1 gene, which acts downstream in the sex determining cascade of mammals, similar to its *Drosophila* and *C. elegans* homologues. In Medaka, the duplication of the chromosomal fragment that included dmrt1a occurred approximately 10 million years ago. After insertion into the proto-Y chromosome a massive degeneration of all genes except dmrt1bY occurred. Subsequent loss of dmrt1bY in several medaka strains reveals the high degree of plasticity of this sex determining mechanism.

From the molecular evolution point of view the main question is how in medaka dmrt1bY could become the master regulator of male development after acquiring an upstream position in the sex-determining cascade. Remarkably, this evolutionary novelty, requiring a rewiring of the regulatory network, was brought about by co-optation of pre-existing cis-regulatory elements contributed by transposable elements. On another level of regulation, differential gene expression of the two dmrt1 co-orthologs seems to be also mediated by post-transcriptional regulation through a highly conserved cis-regulatory motif that directs differential gonadal synexpression of dmrt1 transcripts during gonad development. While the autosomal dmrt1a medaka gene is essential for testis maintenance, in contrast, dmrt1bY was shown to be responsible for male-specific primordial germ cell (PGC) mitotic arrest in the developing gonad at the sex-determination stage, suggesting neo-functionalization.

Funded by

DFG Deutsche
Forschungsgemeinschaft
German Research Foundation

IGB
Leibniz-Institute of
Freshwater Ecology
and Inland Fisheries

Paradigm shift in sex chromosome evolution

Abstracts of the workshop held in Berlin, 19-22 September 2019

Edited by

Matthias Stöck¹  <https://orcid.org/0000-0003-4888-8371>

Lukáš Kratochvíl²  <https://orcid.org/0000-0002-3515-729X>



¹ Leibniz-Institute of Freshwater Ecology and Inland Fisheries (IGB), Department of Ecophysiology and Aquaculture, Berlin, Germany

² Charles University, Faculty of Science, Department of Ecology, Prague, Czech Republic



© 2019. This manuscript version is made available under the [CC-BY 4.0 license](#)