Gudrun Rappold: Selected Research papers

Sex chromosomes, X-inactivation, Recombination

1984	First evidence that closely related sequences on the X and Y chromosome can reside outside the pairing region	Cooke, Brown, Rappold, *Nature,* 1984
1985	Isolation of the first hypervariable sequences from the human sex chromosomes	Cooke, Brown, Rappold, *Nature,* 1985
1987	Identification of a testis-specific gene next to a CpG island	Rappold et al., *EMBO J.*, 1987
1991	First evidence that deletions within the pseudoautosomal region are associated with short stature / linear growth	Henke et al., *Am. J. Hum. Genet.,* 1991
1993	Identification of a pseudoautosomal gene that escapes X-inactivation and its homolog on Xq undergoes X-inactivation	Schiebel et al., *Nature Genet.,* 1993
1994	Double crossover in the human pseudoautosomal region and its bearing on crossover interference	Rappold et al., *Hum. Mol. Genet.,* 1994
1995	Identification of the protein kinase gene PKX1 as a site of chromosomal instability	Klink et al., *Hum. Mol. Genet.,* 1995
1996	Double strand breaks during yeast meiosis reflect meiotic recombination in the human genome	Klein et al., *Nature Genet.,* 1996
1997	Evidence that abnormal XY interchange between protein kinase gene PRKX / PRKY accounts for XX males and XY females	Schiebel et al., *Hum. Mol. Genet.,* 1997
2005	Interchromosomal segmental duplication in the pericentromeric region of human Y chromosome	Kirsch et al., *Genome Res.,* 2005

Developmental homeobox genes *SHOX* and *SHOX2*

1997	Identification of a homeobox gene, S*HOX*, underlying idiopathic short stature and Turner syndrome	Rao et al., *Nature Genet.,* 1997
1998	Identification and functional characterization of the homeobox gene *SHOX2* implicated in brain, heart and limb development	Blaschke et al., *PNAS,* 1998
2000	Evidence that the homeobox gene *SHOX* is involved in skeletal abnormalities in Turner syndrome	Clement-Jones et al., *Hum. Mol. Genet.,* 2000
2001	Evidence that the Léri-Weill and Turner syndrome *SHOX* gene is a cell-type specific activator	Rao et al., *Hum. Mol. Genet.,* 2001
2004	First evidence that *SHOX* induces cellular growth arrest and apoptosis	Marchini et al., *J. Biol. Chem.,* 2004
2007	*SHOX* Enhancer sequences regulate expression in developing chicken limbs and associate with short stature phenotype	Sabherwal et al., *Hum. Mol. Genet*., 2007
2007	First *SHOX2* knock-out mouse model reveals essential functions in pacemaking development	Blaschke et al., *Circulation,* 2007
2007	Isolation and characterization of *BNP* as first transcriptional target of SHOX	Marchini et al., *Hum. Mol. Genet.,* 2007
2010	Evidence that *SHOX2* mediates *Tbx5* activity by regulating *BMP4* in the developing heart	Puskaric et al., *Hum. Mol. Genet.,* 2010
2013	Height matters – from monogenetics to common disease	Durand & Rappold, *Nature Rev. Endocrin.,* 2013
2016	Coding and non-coding *SHOX2* variants in early-onset atrial fibrillation	Hoffmann et al., *Basic Res. Cardiol,* 2016
2016	A track record on *SHOX*: from basic research to complex models and therapy	Marchini, Ogata, Rappold, *Endocrine Rev.,* 2016
2017	Retinoic acid catabolizing enzyme *CYP26C1* is a genetic modifier for severity in *SHOX* deficiency	Montalbano et al., *EMBO Mol. Medicine,* 2017
2020	Precise correction of SHOX2 mutations in iPSCs via gene editing and sib-selection	Sumer, Hoffmann et al., *Stem Cell Reports*, 2020

Neurodevelopmental Disorders: *SHANK*s, *FOXP1* and other disease genes

2000	First link between *VCX* gene and mental retardation	Fukami et al., *Am. J. Hum. Genet.,* 2000
2002	Identification of Rho-GTPase *srGAP3* underlying mental retardation	Endris et al., *PNAS,* 2002
2003	Mutations in *PROSIT240*, a novel *TRAP240*-like gene, in patients with congenital heart defect and intellectual disability	Muncke et al., *Circulation,* 2003
2010	Identification of mutations in the *SHANK2* synaptic scaffolding gene in autism and mental retardation (F1000)	Berkel et al., *Nature Genet.,* 2010
2010	Mutation in *GRIN2A* and *B* encoding regulatory subunits of NMDA receptor cause neurodevelopmental phenotypes	Endele et al., *Nature Genet.,* 2010
2011	Evidence that SrGAP3 interacts with Lamellipodin and regulates Rac-dependent cellular protrusion	Endris et al., *J. Cell Science*, 2011
2012	Genetic mutation associated with intellectual disability: an exome-sequencing study	Rauch et al., *Lancet,* 2012
2012	*SHANK1* Deletions in Males with Autism Spectrum Disorder	Sato et al., *Am.J. Hum. Genet.*, 2012
2012	Inherited and *de novo SHANK2* variants associated with autism spectrum disorder impair neuronal morphogenesis and physiology	Berkel et al., *Hum. Mol. Genet.*, 2012
2014	Genetic and clinical diversity of *SHANK* mutations in autism spectrum disorder	Leblond et al., *PloS Genet*., 2014
2015	Brain-specific *Foxp1* deletion impairs neuronal development and causes autistic behaviour	Bacon et al., *Mol. Psychiat*., 2015
2015	Identification and functional characterisation of *SHANK2* variants in schizophrenia	Peykov et al., *Mol. Psychiat*., 2015
2017	Sex-specific Foxp1 and Foxp2 expression in mouse brain and impact on ultrasonic vocalisation	Fröhlich et al., *Hum. Mol. Genet.,* 2017
2019	Gastrointestinal dysfunction in autism displayed by altered motility and achalasia in *Foxp1*$^{+/-}$ mice	Fröhlich et al., *PNAS,* 2019
2020	Gene mutations driving brain and gut dysfunction in autism	Niesler & Rappold, *Mol. Psychiat*., 2021
2021	Imbalanced pre- and postsynaptic Shank2 functions affect social behaviour	Eltokhi et al., *Mol. Psychiat.,* 2021
2022	Mitochondrial dysfunction and oxidative stress contribute to cognitive and muscle impairment in FOXP1 syndrome	Wang, Fröhlich et al., *PNAS,* 2022

Other topics

1995	Mutation in sulfatase genes on Xp 22.3 in Chondrodysplasia punctata and implication for Warfarin embryopathy	Franco et al., *Cell* 1995
1998	Molecular identification of the corticosterone-sensitive extraneuronal catecholamine transporter	Gründemann et al., *Nature Neuroscience,* 1998
2015	Exploring the genetics of irritable bowel syndrome: a GWA study	Ek et al., *Gut*, 2015
2017	miR-16 and miR-125b are involved in barrier function dysregulation	Martinez et al., *Gut*, 2017
2017	Clinical practice guidelines for the care of girls and women with Turner syndrome	Gravholt et al., *Eur. J. Endocrinol*, 2017
2018	Identification of SLC20A1 and SLC15A4 as risk factors for combined pituitary hormone deficiency	Simm et al, *Genet Med.,* 2018
2020	Inhibition of HDAC6 activity protects dopaminergic neurons from alpha-synuclein toxicity in Parkinson disease	Francelle, Onteiro, Rappold, *Sci Rep,* 2020
2021	Past and present DNA collections from vulnerable groups need highest ethical standards	Lipphardt, ... Rappold, *Nature*, 2021