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
2014	*SHOX* triggers the lysomal pathway of apoptosis via oxidative stress	Hristov et al., *Hum. Mol. Genet.,* 2014
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	Missense mutations and gene interruption in *PROSIT240*, a novel *TRAP240*-like gene, in patients with congenital heart defect and mental retardation	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*., 2020
2021	Imbalanced pre- and postsynaptic Shank2 functions affect social behaviour	Eltokhi et al., 2021
2021	Mitochondrial dysfunction and oxidative stress explain cognitive and muscle impairment in FOXP1 syndrome	Wang, Fröhlich et al., 2021

Other disease 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	Francelle, Onteiro, Rappold, *Sci Rep,* 2020