

# Turner Syndrome

Pathophysiology, Diagnosis  
and Treatment

Patricia Y. Fechner  
*Editor*



Springer

# Turner Syndrome

Patricia Y. Fechner  
Editor

# Turner Syndrome

Pathophysiology, Diagnosis and Treatment

 Springer

*Editor*

Patricia Y. Fechner  
Division of Endocrinology  
Seattle Children's Hospital  
University of Washington  
Seattle, WA  
USA

ISBN 978-3-030-34148-0      ISBN 978-3-030-34150-3 (eBook)  
<https://doi.org/10.1007/978-3-030-34150-3>

© Springer Nature Switzerland AG 2020

This work is subject to copyright. All rights are reserved by the Publisher, whether the whole or part of the material is concerned, specifically the rights of translation, reprinting, reuse of illustrations, recitation, broadcasting, reproduction on microfilms or in any other physical way, and transmission or information storage and retrieval, electronic adaptation, computer software, or by similar or dissimilar methodology now known or hereafter developed.

The use of general descriptive names, registered names, trademarks, service marks, etc. in this publication does not imply, even in the absence of a specific statement, that such names are exempt from the relevant protective laws and regulations and therefore free for general use.

The publisher, the authors and the editors are safe to assume that the advice and information in this book are believed to be true and accurate at the date of publication. Neither the publisher nor the authors or the editors give a warranty, expressed or implied, with respect to the material contained herein or for any errors or omissions that may have been made. The publisher remains neutral with regard to jurisdictional claims in published maps and institutional affiliations.

This Springer imprint is published by the registered company Springer Nature Switzerland AG  
The registered company address is: Gewerbestrasse 11, 6330 Cham, Switzerland

*To my parents, thank you for all of your love  
and support.*

# Preface

The purpose of this book is to provide a resource for providers who care for girls and women with Turner syndrome. Turner syndrome occurs in 1:1800–2000 females, yet many providers may not have a lot of experience caring for a female with Turner syndrome. Patients with Turner syndrome have many of the same health issues as other females, but some health issues are at a higher frequency that necessitates more frequent and earlier screening.

Many contributors to this work are experts in the care of females with Turner syndrome, while others are experts in their own field of medicine who can apply this expertise to the care of individuals with Turner syndrome. They all have worked on this project as a labor of love and for the benefit of the girl or woman with Turner syndrome. I wish to thank each of them for their significant contributions to this book.

The chapters in the book are organized by subspecialty and/or major health issue, with the goal to provide in-depth coverage of the health issues faced by those with Turner syndrome.

Seattle, WA, USA

Patricia Y. Fechner, MD

# Contents

<b>1</b>	<b>Description of Turner Syndrome</b> . . . . .	<b>1</b>
	Alissa J. Roberts and Patricia Y. Fechner	
<b>2</b>	<b>The Genetics of Turner Syndrome</b> . . . . .	<b>13</b>
	Margaret P. Adam and Melanie A. Manning	
<b>3</b>	<b>Pattern and Etiology of Growth Disturbance in Turner Syndrome and Outcomes of Growth-Promoting Treatments</b> . . . . .	<b>33</b>
	Charmian A. Quigley	
<b>4</b>	<b>Fertility Preservation for Turner Syndrome</b> . . . . .	<b>79</b>
	Courtney Finlayson, Lia Bernardi, and Reema Habiby	
<b>5</b>	<b>Estrogen Replacement in Turner Syndrome</b> . . . . .	<b>93</b>
	Karen O. Klein, Robert L. Rosenfield, Richard J. Santen, Aneta M. Gawlik, Philippe Backeljauw, Claus H. Gravholt, Theo C. J. Sas, and Nelly Maura	
<b>6</b>	<b>The Heart and Vasculature in Turner Syndrome: Development, Surveillance, and Management</b> . . . . .	<b>123</b>
	Luciana T. Young and Michael Silberbach	
<b>7</b>	<b>Renal Disorders and Systemic Hypertension</b> . . . . .	<b>139</b>
	Yosuke Miyashita and Joseph T. Flynn	
<b>8</b>	<b>Endocrine and Metabolic Consequences of Turner Syndrome</b> . . . . .	<b>157</b>
	Mette H. Viuff and Claus H. Gravholt	
<b>9</b>	<b>Care of Girls with Turner Syndrome: Beyond Growth and Hormones</b> . . . . .	<b>175</b>
	Angel Siu Ying Nip and Darcy King	
<b>10</b>	<b>Ear and Hearing Problems in Turner Syndrome</b> . . . . .	<b>185</b>
	Åsa Bonnard and Malou Hultcrantz	

**11 Ocular Features in Turner Syndrome** ..... 199  
Erin P. Herlihy and Jolene C. Rudell

**12 Gastrointestinal and Hepatic Issues in Women with Turner Syndrome** ..... 205  
Ghassan T. Wahbeh, Amanda Bradshaw, Lauren White, and Dale Lee

**13 Dermatologic Conditions in Turner Syndrome** ..... 221  
Alessandra Haskin and Eve Lowenstein

**14 Orthopedic Manifestations in Turner Syndrome** ..... 237  
Anna M. Acosta, Suzanne E. Steinman, and Klane K. White

**15 Oral Manifestations in Turner Syndrome** ..... 249  
Carolina Di Blasi and Harlyn Susarla

**16 The Turner Syndrome Resource Center: An Interdisciplinary Approach to the Care of Girls and Women with Turner Syndrome** ..... 257  
Philippe Backeljauw and Sarah Corathers

**17 Future Directions** ..... 267  
Patricia Y. Fechner

**Index** ..... 271



# Contributors

**Anna M. Acosta, MD** Memorial Care and Miller Children's and Women's Hospital, Long Beach, CA, USA

**Margaret P. Adam, MD** Division of Genetic Medicine, Department of Pediatrics, University of Washington School of Medicine and Seattle Children's Hospital, Seattle, WA, USA

**Philippe Backeljauw, MD** The Cincinnati Center for Pediatric and Adult Turner Syndrome Care, Cincinnati Children's Hospital Medical Center, University of Cincinnati College of Medicine, Cincinnati, OH, USA

**Lia Bernardi, MD, MSCI** Division of Reproductive Endocrinology, Department of Obstetrics and Gynecology, Northwestern University Feinberg School of Medicine, Chicago, IL, USA

**Carolina Di Blasi, MD** Seattle Childrens Hospital, University of Washington, Seattle, WA, USA

**Åsa Bonnard, MD, PhD** Department of Clinical Science, Intervention and Technology, Division of Otorhinolaryngology, Karolinska Institutet, Stockholm, Sweden

Department of Otorhinolaryngology, Karolinska University Hospital, Stockholm, Sweden

**Amanda Bradshaw, PA-C** Gastroenterology, Seattle Children's Hospital, Seattle, WA, USA

**Sarah Corathers, MD** The Cincinnati Center for Pediatric and Adult Turner Syndrome Care, Cincinnati Children's Hospital Medical Center, University of Cincinnati College of Medicine, Cincinnati, OH, USA

**Patricia Y. Fechner, MD** Division of Endocrinology, Seattle Children's Hospital, University of Washington, Seattle, WA, USA

**Courtney Finlayson, MD** Division of Endocrinology, Department of Pediatrics, Ann & Robert H. Lurie Children's Hospital of Chicago, Northwestern University Feinberg School of Medicine, Chicago, IL, USA

**Joseph T. Flynn, MD, MS** University of Washington School of Medicine, Seattle, WA, USA

Division of Nephrology, Seattle Children's Hospital, Seattle, WA, USA

**Aneta M. Gawlik, PhD** Department of Pediatrics and Pediatric Endocrinology, School of Medicine in Katowice, Medical University of Silesia, Katowice, Poland

**Claus H. Gravholt, MD, PhD** Department of Endocrinology and Internal Medicine, Aarhus University Hospital, Aarhus, Denmark

Department of Molecular Medicine, Aarhus University Hospital, Aarhus, Denmark

Department of Endocrinology and Internal Medicine and Medical Research Laboratories, Aarhus University Hospital, Aarhus, Denmark

**Reema Habiby, MD** Division of Endocrinology, Department of Pediatrics, Ann & Robert H. Lurie Children's Hospital of Chicago, Northwestern University Feinberg School of Medicine, Chicago, IL, USA

**Alessandra Haskin, BA** Department of Dermatology, SUNY Health Science Center at Brooklyn, Brooklyn, NY, USA

**Erin P. Herlihy, MD** Department of Ophthalmology, University of Washington School of Medicine, Seattle, WA, USA

Division of Pediatric Ophthalmology, Seattle Children's Hospital, Seattle, WA, USA

**Malou Hultcrantz, MD** Department of Clinical Science, Intervention and Technology, Division of Otorhinolaryngology, Karolinska Institutet, Stockholm, Sweden

Department of Otorhinolaryngology, Karolinska University Hospital, Stockholm, Sweden

**Darcy King, ARNP** University of Washington, Seattle, WA, USA

**Karen O. Klein, MD** University of California, San Diego & Rady Children's Hospital, San Diego, CA, USA

**Dale Lee, MD** Pediatrics-Gastroenterology, Clinical Nutrition & Celiac Program, Seattle Children's Hospital, University of Washington, Seattle, WA, USA

**Eve Lowenstein, MD, PhD** Department of Dermatology, SUNY Health Science Center at Brooklyn, Brooklyn, NY, USA

**Melanie A. Manning, MD** Division of Medical Genetics, Department of Pediatrics, Stanford University School of Medicine and Lucile Salter Packard Children's Hospital, Stanford, CA, USA

Department of Pathology, Stanford University School of Medicine, Stanford, CA, USA

**Nelly Mauras, MD** Nemours Children's Health System, Jacksonville, FL, USA