



**FIGURE 45-8** Hypoplastic left leg and distal digital hypoplasia in an infant delivered by cesarean section owing to extensive uterine leiomyomata, which were so enlarged that they had to be surgically resected so that the baby could be delivered.

micrognathia, and clubfoot), which is similar to the 30% risk suggested previously.<sup>1</sup> Vaginal bleeding was significantly more common than in mothers with a normal uterus (54.1% versus 14.1%), and vaginal bleeding has previously been associated with limb reduction defects.<sup>13,14,16</sup> In a study of 322 women with abnormal uterine bleeding, hysteroscopy detected asymptomatic Müllerian anomalies in 10% of these women. The women with Müllerian anomalies had a significantly higher incidence of spontaneous abortion and lower cumulative live birth rates,<sup>13,17</sup> which suggests that some fetuses with severe defects might have been lost earlier in gestation in this case-control study of liveborn and stillborn infants.<sup>18</sup>

## MANAGEMENT, PROGNOSIS, AND COUNSEL

Surgical improvement of the uterine size, if indicated and possible, may improve the chances of rearing a normal fetus to a term birth.<sup>11</sup> Large uterine fibroids may also merit consideration of surgical intervention. Among 174 patients with appropriate imaging, reproductive performance depended on the type of uterine malformation and urinary

tract anomalies. The lowest percentage of women who had only live births occurred in women with bicornuate unicornis uterus (28%). Among women with uterine anomalies and unilateral renal agenesis, perinatal outcomes were much better (72% had only living children) than those with no renal agenesis (40%). The 355 pregnancies in patients with associated unilateral renal agenesis were associated with term deliveries and living children, while pregnancies in women with uterine malformation and no renal agenesis had more fetal loss, premature birth, and breech presentations. Thus if the Müllerian anomaly was the consequence of abnormal mesonephric or Wolffian morphogenesis, reproductive performance was much improved over women with an isolated Müllerian defect.<sup>19</sup> Among 21 women with uterine malformations and a surgically treated obstructed hemivagina and associated ipsilateral renal agenesis (12 didelphic, 6 separate, and 3 bicornuate uterus), all had surgical excision of the longitudinal vaginal septum causing the obstructed hemivagina during adolescence. Conception was attempted at a median of 13 years later in 13 of these women, who produced 22 pregnancies, with 77% of pregnancies occurring contralateral to the treated obstructed hemivagina and unilateral renal agenesis, and 91% of these pregnancies ended in

# Smiths Recognizable Patterns Of Human Deformation

**Léo Pomar**



## **Smiths Recognizable Patterns Of Human Deformation:**

Smith's Recognizable Patterns of Human Malformation Kenneth Lyons Jones, 1997 This user friendly 5th Edition provides concise but complete information on numerous common and rare disorders that cause human malformation Includes an outline of the salient features of each condition as well as material on natural history etiology and pathogenesis The text is accompanied by helpful illustrations and reference lists Organized to allow for easy access to essential information

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*Syndromes of the Head and Neck* Robert J. Gorlin, M. Michael Cohen Jr., D.M.D., Ph.D., Raoul C.M. Hennekam M.D., Ph.D., 2001-09-27 This classic text one of the true anchors of our clinical genetics publishing program covers over 700 different genetic syndromes involving the head and neck and it has established itself as the definitive comprehensive work on the subject The discussion covers the phenotype spectrum epidemiology mode of inheritance pathogenesis and clinical profile of each condition all of which is accompanied by a wealth of illustrations The authors are recognized leaders in the field and their vast knowledge and strong clinical judgment will help readers make sense of this complex and burgeoning field Dr Gorlin retires as editor in this edition and co editor Raoul Hennekam takes over Dr Hennekam is regarded as one of the top dysmorphologists and indeed one of the top clinical geneticists in the world Judith Allanson is new to the book but is a veteran OUP author and a widely respected geneticist and Ian Krantz at Penn is a rising star in the field Dr Gorlin's name has always been closely associated with the book and it has now become part of the title As in all fields of genetics there has been an explosion in the genetics of dysmorphology syndromes and the author has undertaken a complete updating of all chapters in light of the discoveries of the Human Genome Project and other ongoing advances with some chapters requiring complete rewriting Additional material has been added both in terms of new syndromes and in updating information on existing syndromes The book will appeal to clinical geneticists pediatricians neurologists head and neck surgeons otolaryngologists and dentists The 4th edition which published in 2001 has sold 2 600 copies      **Human Malformations and Related Anomalies** Roger E. Stevenson, 2016 The central theme of this text is to provide information on individual anomalies et to connect these anomalies to the malformation syndromes et associated problems primarily through the use of differential diagnostic tables      Emery and Rimoin's Essential Medical Genetics David L. Rimoin, Reed E. Pyeritz, Bruce Korf, 2013-05-15 For decades Emery and Rimoin's Principles and Practice of Medical Genetics has provided the ultimate source for practicing

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**Color Doppler, 3D and 4D Ultrasound in Gynecology, Infertility and Obstetrics** Sanja Kupesic Plavsic, 2014-05-14 Doody Rating 3 stars Over the last decade impressive improvements in computer and ultrasound technology have promoted a wide use of ultrasound in clinical practice With the advent of color and power Doppler ultrasound and more recently three 3D and four dimensional 4D ultrasound research expansion in the field of human reproduction obstetrics and gynecologic oncology has occurred Ultrasound has simplified guided techniques such as oocyte collection and breast biopsy but has also become an important technique in the assessment of the follicular growth and endometrial development as well as in evaluation of the uterine and ovarian perfusion Significant studies have been made in the gynecological application of Doppler sonography and screening for ovarian and uterine malignancy In obstetrics Doppler sonography has allowed unprecedented insight in the pathophysiology of human fetal development In a relatively short period of time 3D and 4D ultrasound has proved to be a useful clinical tool in almost all sections of gynecology and obstetrics In this book the authors explain the significance of each of the discussed subjects in an effective way by integrating important and updated information and illustrative examples The contributors of this edition have made significant improvements included updated

information and a few unique illustrations Each chapter has been reviewed and revised to focus on the clinicians needs in ultrasound practice The educational impact of the book is further enhanced by adding a manual for sonographers and physicians entitled Clinical Sonographic Pearls that was created for better organization of important clinical presentation based information

**Berman's Pediatric Decision Making** Lalit Bajaj, Simon Hambidge, Ann-Christine Nyquist, Gwendolyn Kerby, 2011-06-30 Berman's Pediatric Decision Making uses an algorithmic structured approach to lead you to the right diagnosis and treatment every time Drs Lalit Baja Simon Hambidge Ann Christine Nyquist and Gwendolyn Kerby use evidence based research and flow charts for each presenting complaint or specific disorder to provide quick access to the information you need for effective decision making With updated drug tables revised algorithms and full text online access at [www.expertconsult.com](http://www.expertconsult.com) this streamlined new edition makes it even easier for you to diagnose and manage common clinical problems from infancy through adolescence Rapidly access guidance on diagnosis and management from algorithms for each clinical disorder Treat the full range of diseases and disorders with comprehensive coverage of diagnosis assessment of severity and clinical management Choose the best treatment for each case thanks to indications for surgical interventions as well as expensive diagnostic procedures Access the fully searchable contents online at [www.expertconsult.com](http://www.expertconsult.com) Stay current on recent developments and make effective decisions for movement disorders physical abuse in children sexual abuse in children eating disorders ADHD and other hot topics Find answers quickly and easily with a new table of contents organized into two sections Presenting Complaints and Specific Disorders that reduces the need to flip between chapters Tap into the diverse perspectives of expert authors from all over the country Get only the information you need in the streamlined new edition with shorter more user friendly flow diagrams and fewer specialized chapters

**Syndromes of the Head and Neck, An Issue of Atlas of the Oral & Maxillofacial Surgery Clinics** Dean M. DeLuke, 2014-09-05 This issue of the Atlas of the Oral and Maxillofacial Surgery Clinics of North America serves as an aid to identification of 70 of the more important head and neck syndromes and organized using the same nosology as in the landmark text of Robert Gorlin As such it should be useful to a varied audience including not only the oral and maxillofacial surgeon but also the oral pathologist the practitioner of oral medicine and members of the craniofacial team the orthodontist pediatric dentist speech and language specialist geneticist plastic surgeon ENT specialist pediatrician nurse coordinator and others Areas covered include Craniosynostosis Syndromes Syndromes Affecting Bone Metabolic and Autoimmune Syndromes Syndromes Affecting Skin and Mucosa Hamartoneoplastic Syndromes Branchial Arch Syndromes Clefting Syndromes Syndromes Affecting the Central Nervous System Chromosomal Syndromes Syndromes with Unusual Facies and Syndromes with Unusual Dental Findings or Gingival Components



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