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This revised edition of its critically acclaimed predecessor incorporates new research findings and advances in the treatment of genetic disorders, including gene-directed therapies. Notably, each chapter incorporates thoroughly updated material on new genetic findings, consensus diagnostic criteria, and management strategies.

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Management of Genetic Syndromes From Wiley-Liss Bibliography

- Sales Rank: #3853414 in Books
- Published on: 2004-11-25
- Original language: English
- Number of items: 1
- Dimensions: 11.24" h x 1.63" w x 8.82" l,
- Binding: Hardcover
- 720 pages



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