TABLE OF CONTENTS

1.0 Introduction 22
   1.1 Executive Summary 23
   1.2 Objectives of this Report 25
   1.3 Key Questions Answered in this Report 25

2.0 Prenatal Pregnancy Complications: An Overview 27
   2.1 Advanced Maternal Age (AMA) 27
      2.1.1 Risk of Down Syndrome with Increased AMA 27
      2.1.2 Risk of Miscarriage with Increased AMA 28
      2.1.3 Risk of Cesarean Section with Increasing AMA 29
      2.1.4 Risk of Gestational Diabetes with AMA 30
      2.1.5 Risk for Pregnancy-Induced Hypertension with AMA 31
      2.1.6 Risk of Placenta Previa with AMA 31
      2.1.7 Delayed First Pregnancy in the U.S. 32
         2.1.7.1 First Birth Rates for 35-39 Aged U.S. Women by Race 33
   2.2 Pre-Existing Maternal Medical Conditions 34
      2.2.1 Pregnancy Outcome in Women with Renal Disease 35
      2.2.2 Pregnancy Outcome in Diabetic Women 36
      2.2.3 Pregnancy Outcome in Women with Pre-Existing Thyroid Disease 36
      2.2.4 Pregnancy Outcome in Obese Women 36
      2.2.5 Pregnancy Outcome in Asthmatic Women 37
      2.2.6 Pregnancy Outcome in Women with Epilepsy 38
      2.2.7 Pregnancy Outcome in Women with Autoimmune Diseases 38
      2.2.8 Pregnancy Outcome in Women with Hemoglobinopathies 39
      2.2.9 Pre-Existing Chronic Diseases among Women of Reproductive Ages in the U.S. 40
         2.2.9.1 Chronic Disease Risk Behaviors and Risk Factors among Women of Reproductive Ages in the U.S. 41
   2.3 Medical Conditions Occurring During Pregnancy 42
      2.3.1 Preeclampsia (Toxemia) 42
         2.3.1.1 Prevalence of Preeclampsia in the U.S. 42
      2.3.2 Gestational Diabetes in the U.S. 44
         2.3.2.1 Prevalence of Gestational Diabetes in the U.S. 44
   2.4 Pregnancy-Related Issues 46
      2.4.1 Premature Labor 46
         2.4.1.1 Common Medical Complications in Premature Babies 47
      2.4.1.2 Global Prevalence of Preterm Labor 47
      2.4.1.3 Prevalence of Preterm Births in the U.S. by Stage 48
      2.4.1.4 Prevalence of Preterm Births in the U.S. by Race/Ethnicity 48
      2.4.1.5 Prevalence of Preterm Births in the U.S. by Plurality of Birth 49
      2.4.1.6 Prevalence of Preterm Births in the U.S. by Maternal Age 50
      2.4.1.7 Multiple Births in the U.S. 51
      2.4.2 Placenta Previa 52
      2.4.3 Breech Presentation 52
      2.4.4 Meconium Stained Liquor 52
      2.4.5 Oligohydramnios and Polyhydramnios 53
   2.5 Fetal Problems 54
      2.5.1 Fetal Growth Restriction 54
      2.5.2 Imperforate Anus 54
      2.5.3 Congenital Heart Disease 55

3.0 Types of Genetic Diseases in Fetuses: An Overview 56
   3.1 Single Gene Disorders 56
      3.1.1 Autosomal Dominant Genetic Disorders 56
3.1.1.1 Huntington Disease (HD)  57
3.1.1.2 Familial Hypercholesterolaemia (FH)  58
3.1.1.3 Marfan syndrome  58
3.1.1.4 Myotonic Dystrophy  59
3.1.2 Autosomal Recessive Genetic Disorders  60
3.1.2.1 Cystic Fibrosis (CF)  61
3.1.2.2 Canavan Disease  65
3.1.2.3 Neutropenia  65
3.1.2.4 Ellis-van Creveld Syndrome (EVC)  66
3.1.2.5 Familial Mediterranean fever (FMF)  66
3.1.2.6 Fanconi Anemia (FA)  66
3.1.2.7 Gaucher Disease  67
3.1.2.8 Mucopolysaccharidosis (MPSs)  67
3.1.2.9 Phenylketonuria (PKU)  68
3.1.2.10 Sickle Cell Disease  69
3.1.2.11 Beta-Thalassaemia  69
3.1.2.12 Inheritance Pattern of Autosomal Recessive Genetic Disorders  70
3.1.2.13 Chances of Inheriting a Single Gene Disorder  73
3.1.2.14 Common Recessive Disease Traits in Selected Ethnic Groups  73
3.1.2.15 Comprehensive List of Common Single Gene Disorders  74
3.1.3 X-Linked Dominant Genetic Disorders  75
3.1.3.1 Hypophosphotemic Rickets  76
3.1.3.2 Incontinentia Pigmenti  76
3.1.3.3 Focal Dermal Hypoplasia  76
3.1.3.4 Orofaciodigital Syndrome  77
3.1.3.5 Inheritance of Sex-Linked Dominant Disorders  77
3.1.4 X-Linked Recessive Disorders  78
3.1.4.1 Lesch-Nyhan Syndrome  79
3.1.4.2 Menkes Disease (Kinky hair syndrome)  79
3.1.4.3 Hemophilia A and B  80
3.1.4.4 Fabry’s Disease  80
3.1.4.5 Wiskott-Aldrich syndrome (WAS)  81
3.1.4.6 Bruton’s Agammaglobulinemia  82
3.1.4.7 Color Blindness  82
3.1.4.8 Complete Androgen Insensitivity Syndrome  83
3.1.4.9 Inheritance of X-Linked Recessive Traits  83
3.2 Chromosomal Disorders  85
3.2.1 47, XXY (Klinefelter Syndrome)  86
3.2.1.1 Prenatal Diagnosis of Klinefelter Syndrome  86
3.2.2 47, XYY Syndrome  87
3.2.2.1 Diagnosis of 47, XYY Syndrome  87
3.2.3 45, X Syndrome (Turner Syndrome)  87
3.2.3.1 Prenatal Diagnosis of Turner Syndrome  88
3.2.4 47, XXX (Triple X Syndrome)  88
3.2.4.1 Tests for Triple X Syndrome  89
3.2.5 Trisomy 21 (Down Syndrome)  89
3.2.5.1 Tests for Down Syndrome  89
3.2.5.2 Incidence of Down Syndrome by Maternal Age  90
3.2.5.3 Developmental Delay in Children with Down Syndrome  91
3.2.6 Trisomy 18 (Edward’ s Syndrome)  92
3.2.6.1 Prenatal Diagnosis of Trisomy 18  92
3.2.6.2 Clinical Manifestations of Trisomy 18  92
3.2.7 Trisomy 13 (Patau Syndrome)  93
3.2.7.1 Prenatal Diagnosis of Trisomy 13  94
3.2.8 Triploid Syndrome  94

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3.2.8.1 Congenital Anomalies Associated with Triploidy
3.2.8.2 Prenatal Diagnosis of Triploidy
3.2.9 Prevalence of Trisomies in the U.S.
3.2.9.1 Trisomies and Abortions
3.2.10 Maternal Age-Related Frequencies of Aneuploid Fetuses
3.2.11 Clinical Features of Common Chromosomal Aneuploidy
3.2.12 Maternal Age and Chromosomal Aneuploidy
3.2.13 Sensitivity and Specificity of Maternal Serum and Ultrasound Tests for Aneuploids
3.3 Structural Chromosomal Abnormalities
3.3.1 Autosomal Deletions
3.3.1.1 Wolf-Hirschhorn Syndrome (WHS)
3.3.1.2 Cri du chat Syndrome
3.3.1.3 Langer-Giedion Syndrome
3.3.2 Common Autosomal Microdeletion Syndrome
3.3.2.1 Williams Syndrome
3.3.2.2 WAGR Syndrome
3.3.2.3 Prader-Willi Syndrome (PWS)
3.3.2.4 Angelman Syndrome
3.3.2.5 Miller-Dieker Syndrome
3.3.2.6 Smith-Magenis Syndrome (SMS)
3.3.2.7 Alagille Syndrome (ALGS)
3.3.2.8 DiGeorge Syndrome
3.3.3 Autosomal Duplication Syndromes
3.3.3.1 Beckwith-Wiedemann Syndrome (BWS)
3.3.3.2 Charcot-Marie-Tooth Disease Type 1A (CMT1A)
3.3.3.3 Cat-Eye Syndrome
3.3.4 Appropriate Technologies for the Detection of Microdeletions, Duplications and Copy Number Variants

4.0 Genetic Counseling: An Overview
4.1 Impact of Recent Advances in Clinical Genomics on Genetic Counseling
4.2 Genetic Counselors in DTC GT Industry
4.2.1 Types of Genetic Counseling Offered by DTC GT Companies
4.2.2 Roles of Genetic Counselors in DCT GT Industry

5.0 Prenatal Screening for Genetic Diseases: An Overview
5.1 Routine Prenatal Screening Tests
5.2 Less-Routine Prenatal Screening Tests
5.3 Beta Human Chorionic Gonadotropin (β-hCG) Screening Test
5.3.1 hCG Kits and Manufacturers
5.4 First Trimester Pregnancy-Associated Plasma Protein-A (PAPP-A) Screening Test
5.5 Maternal Serum Alpha-Fetoprotein (MSAFP) Screening Test
5.6 Second Trimester Serum β-hCG Screening Test
5.7 Second Trimester Unconjugated Estriol (uE3) Screening Test
5.8 Second Trimester Inhibin-A Screening Test
5.9 Second Trimester Hexosaminidase Test
5.10 Second Trimester Triple-Screen Quad Screen Tests
5.11 Second Trimester Screening of Cell-Free Fetal DNA (cffDNA)
5.11.1 Sensitivity and Specificity of NIPTs
5.11.2 Diagnostic Applications of cffDNA
5.11.3 Time of Availability of cffDNA in Maternal Blood
5.11.4 The Cost of NIPTs
5.11.5 Cost of NIPTs by Product
5.11.6 History of Aneuploidy Testing from 1970s to 2011
5.11.7 NIPT Methods of Detecting Aneuploidy
5.11.7.1 Shotgun Method

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<table>
<thead>
<tr>
<th>Section</th>
<th>Title</th>
<th>Page</th>
</tr>
</thead>
<tbody>
<tr>
<td>5.11.7.2</td>
<td>Targeted Massively Parallel Sequencing (t-MPS)</td>
<td>126</td>
</tr>
<tr>
<td>5.11.7.3</td>
<td>Single Nucleotide Polymorphisms (SNPs)</td>
<td>126</td>
</tr>
<tr>
<td>5.11.8</td>
<td>Comparison of Amniocentesis and NIPT</td>
<td>127</td>
</tr>
<tr>
<td>5.11.9</td>
<td>Advantages of NIPTs</td>
<td>127</td>
</tr>
<tr>
<td>5.11.10</td>
<td>Disadvantages of NIPTs</td>
<td>128</td>
</tr>
<tr>
<td>5.11.11</td>
<td>Comparison of Performance Criteria for Commonly used Prenatal Tests</td>
<td>128</td>
</tr>
<tr>
<td>5.11.12</td>
<td>Comparison of NIPT Detection Rates with Traditional Tests</td>
<td>129</td>
</tr>
<tr>
<td>5.11.13</td>
<td>False Positive Rates of NIPTs Compared with Those of Traditional Tests</td>
<td>129</td>
</tr>
<tr>
<td>5.11.14</td>
<td>Fetal Genetic Disorders Detected by NIPTs and other Screening Tests</td>
<td>130</td>
</tr>
<tr>
<td>5.12</td>
<td>First Trimester Ultrasound Screening Test</td>
<td>130</td>
</tr>
<tr>
<td>5.12.1</td>
<td>Nuchal Translucency (NT) Screening Test</td>
<td>131</td>
</tr>
<tr>
<td>5.13</td>
<td>Magnetic Resonance Imaging (MRI) in Prenatal Screening</td>
<td>132</td>
</tr>
<tr>
<td>5.14</td>
<td>Prenatal Diagnosis of Genetic Disorders</td>
<td>132</td>
</tr>
<tr>
<td>5.14.1</td>
<td>Amniocentesis (AC)</td>
<td>133</td>
</tr>
<tr>
<td>5.14.2</td>
<td>Reliability of Amniocentesis</td>
<td>133</td>
</tr>
<tr>
<td>5.14.2.1</td>
<td>Reliability of CVS Test</td>
<td>134</td>
</tr>
<tr>
<td>5.14.3</td>
<td>Cordocentesis or Percutaneous Umbilical Blood Sampling</td>
<td>135</td>
</tr>
<tr>
<td>5.15</td>
<td>Advantages and Disadvantages of Prenatal Diagnostic Tests</td>
<td>135</td>
</tr>
<tr>
<td>5.16</td>
<td>Future of Invasive Prenatal Diagnostic Tests</td>
<td>136</td>
</tr>
<tr>
<td>5.16.1</td>
<td>Indispensability of Invasive Diagnostic Tests</td>
<td>137</td>
</tr>
<tr>
<td>5.17</td>
<td>Intact Circulating Fetal Cells for Noninvasive Prenatal Tests</td>
<td>137</td>
</tr>
<tr>
<td>5.17.1</td>
<td>Scheme for Isolating Intact Fetal Cells from Maternal Blood</td>
<td>139</td>
</tr>
<tr>
<td>5.18</td>
<td>Trophoblast Retrieval and Isolation from Cervix (TRIC)</td>
<td>140</td>
</tr>
</tbody>
</table>

### Newborn Screening

<table>
<thead>
<tr>
<th>Section</th>
<th>Title</th>
<th>Page</th>
</tr>
</thead>
<tbody>
<tr>
<td>6.1</td>
<td>Status of Newborn Screening in Developed and Developing Countries</td>
<td>142</td>
</tr>
<tr>
<td>6.2</td>
<td>Status of Newborn Screening in Middle East and North Africa (MENA)</td>
<td>142</td>
</tr>
<tr>
<td>6.3</td>
<td>Screening Tests Recommended for Newborns in the U.S.</td>
<td>143</td>
</tr>
<tr>
<td>6.4</td>
<td>Most Common Newborn Genetic Disorders</td>
<td>145</td>
</tr>
<tr>
<td>6.4.1</td>
<td>Phenylketonuria (PKU)</td>
<td>145</td>
</tr>
<tr>
<td>6.4.2</td>
<td>Screening Tests Used for Diagnosis of PKU</td>
<td>146</td>
</tr>
<tr>
<td>6.4.2.1</td>
<td>Inheritance of CHT</td>
<td>146</td>
</tr>
<tr>
<td>6.4.2.2</td>
<td>Signs and Symptoms of CHC</td>
<td>146</td>
</tr>
<tr>
<td>6.4.2.3</td>
<td>Diagnosis of CHT</td>
<td>146</td>
</tr>
<tr>
<td>6.4.3</td>
<td>Congenital Adrenal Hyperplasia</td>
<td>147</td>
</tr>
<tr>
<td>6.4.3.1</td>
<td>Inheritance of Congenital Adrenal Hyperplasia</td>
<td>147</td>
</tr>
<tr>
<td>6.4.3.2</td>
<td>Symptoms of Congenital Adrenal Hyperplasia</td>
<td>147</td>
</tr>
<tr>
<td>6.4.3.3</td>
<td>Diagnosis of Congenital Adrenal Hyperplasia</td>
<td>147</td>
</tr>
<tr>
<td>6.4.4</td>
<td>Galactosemia</td>
<td>147</td>
</tr>
<tr>
<td>6.4.4.1</td>
<td>Signs and Symptoms of Galactosemia</td>
<td>148</td>
</tr>
<tr>
<td>6.4.4.2</td>
<td>Diagnosis of Galactosemia</td>
<td>148</td>
</tr>
<tr>
<td>6.4.5</td>
<td>Sickle Cell Disease (SCD)</td>
<td>148</td>
</tr>
<tr>
<td>6.4.5.1</td>
<td>Signs and Symptoms of SCD</td>
<td>148</td>
</tr>
<tr>
<td>6.4.5.2</td>
<td>Diagnosis of SCD</td>
<td>148</td>
</tr>
<tr>
<td>6.4.6</td>
<td>Biotidinase Deficiency</td>
<td>149</td>
</tr>
<tr>
<td>6.4.6.1</td>
<td>Inheritance of Biotidinase Deficiency</td>
<td>149</td>
</tr>
<tr>
<td>6.4.6.2</td>
<td>Signs and Symptoms of Biotidinase Deficiency</td>
<td>149</td>
</tr>
<tr>
<td>6.4.6.3</td>
<td>Diagnosis of Biotidinase Deficiency</td>
<td>149</td>
</tr>
<tr>
<td>6.4.7</td>
<td>Homocystinuria</td>
<td>150</td>
</tr>
<tr>
<td>6.4.7.1</td>
<td>Signs and Symptoms of Homocystinuria</td>
<td>150</td>
</tr>
<tr>
<td>6.4.7.2</td>
<td>Diagnosis of Homocystinuria</td>
<td>151</td>
</tr>
<tr>
<td>6.4.8</td>
<td>Maple Syrup Urine Disease (MSUD)</td>
<td>151</td>
</tr>
<tr>
<td>6.4.8.1</td>
<td>Inheritance of MSUD</td>
<td>151</td>
</tr>
<tr>
<td>6.4.8.2</td>
<td>Diagnosis of MSUD</td>
<td>151</td>
</tr>
</tbody>
</table>
7.0 Preimplantation Genetic Screening & Genetic Diagnosis (PGS/PGD) 152
7.1 PGS/PGD Technologies 152
7.1.1 Fluorescent in situ Hybridization (FISH) 152
7.1.2 Array Comparative Genome Hybridization (aCGH) 153
7.1.3 Single Nucleotide Polymorphism (SNP) Microarray 153
7.1.4 Oligonucleotide Genome Sequencing (OGS) 153
7.1.5 Express Genome Sequencing (EGS) 153
7.1.6 Next Generation Sequencing (NGS) 153
7.2 Strengths and Weaknesses of Currently Available PGS/PGD Technologies 153
7.3 Genetic Diseases Detected During PGD 154
7.4 PGS/PGD Testing Products in the Market 159
7.5 Cost of in vitro Fertilization and Related Procedures 160
8.0 Pregnancy, Prenatal, Newborn and PGD-Related Technologies: An Overview 162
8.1 Beta Human Chorionic Gonadotropin (β-hCG) Test 162
8.1.1 Setting up the Test Strip 163
8.1.2 Procedure of the Test 163
8.1.3 Interpretation of β-hCG Test Result 163
8.2 Pregnancy Associated Plasma Protein-A (PAPP-A) Test 164
8.2.1 Principle of PAPP-A Test 164
8.3 Maternal Serum Alpha Fetoprotein (MSAFP) Test 164
8.3.1 Principle of MSAFP Test 164
8.4 Unconjugated Estriol (uE3) Test 165
8.4.1 Principle of uE3 Test 165
8.5 Inhibin A Test 165
8.5.1 Principle of Inhibin A Test 165
8.6 Fetal Karyotyping 166
8.7 Extended Banding Chromosome Studies 166
8.8 Innovation in Invasive Prenatal Diagnosis 166
8.8.1 Microarray: An Alternative for Karyotyping 167
8.8.2 Rapid Aneuploid Detection by Quantitative Fluorescent PCR (QF-PCR) 167
8.8.2.1 Principles of QF-PCR 167
8.8.3 Fluorescence in situ Hybridization (FISH) 168
8.8.3.1 The Format of Fish Test Result 169
8.8.3.2 Commonly Used FISH-Based Tests 170
8.8.3.3 Microdeletions/Microduplications Detectable by FISH 171
8.8.3.4 Types of FISH Probes and Their Functions 171
8.8.4 Microarray-Based Comparative Genomic Hybridization (array-CGH) 172
8.9 Advances in Prenatal Screening: The Power to Know Sooner 173
8.9.1 NIPTs: An Overview of Tests and Technologies 174
8.9.1.1 Harmony Test 174
8.9.1.2 InformaSeq 176
8.9.1.3 Panorama Test 176
8.9.1.4 Prena Test 177
8.9.1.5 NIFTY Test 178
8.9.1.6 IONA Test 178
8.9.1.7 Verifi Test 180
8.9.1.8 MaterniT GENOME Test 181
8.9.1.9 MaterniT21 PLUS Test 182
8.9.1.10 HerediT UNIVERSAL Carrier Screen 183
8.9.2 Comparison of MaterniT21 PLUS, Verifi, Harmony and Panorama Tests 183
8.9.3 U.S. Patents for NIPTs by Company 184
8.9.3.1 Selected Issued Patents of NIPTs 184
8.9.3.2 Assignees of Major U.S. Patents and their Current Licensees 185
8.9.4 Global Availability of NIPTs 186
8.9.5 Cost Effectiveness of NIPTs 187

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GLOBAL PRENATAL & MATERNAL DIAGNOSTIC MARKET TO 2020

8.9.6 Clinical Implementation Strategies to be adopted for NIPTs 188
8.9.7 Integration of NIPT into Healthcare System: Patient-Directed Model 189
8.10 Tandem Mass Spectrometry (MS/MS) in Newborn Screening 190
8.11 Fetal Ultrasound in Prenatal Screening 191
8.11.1 Types of Fetal Ultrasound Exams 191
8.11.2 Best Obstetric and Gynecological Ultrasound Systems in the Market 192
8.11.3 Best Premium Fetal Ultrasound Machines 192
8.11.3.1 Philips Epiq 7 192
8.11.3.2 GE Voluson E8 193
8.11.3.3 GE Voluson E10 193
8.11.3.4 Samsung UGEO WS80A 194
8.11.4 High-End Systems 194
8.11.4.1 Philips Epiq 5 195
8.11.4.2 Philips Affinity 70 195
8.11.4.3 GE Voluson E6 195
8.11.4.4 GE Voluson S8196 196
8.11.5 Midrange Systems 196
8.11.5.1 Philips Affinity 50 197
8.11.5.2 GE Voluson S6197 198
8.11.5.3 Samsung AccuVix A30197 198
8.11.5.4 Philips Clear Vue 650 198
8.11.6 Economy Systems 198
8.11.6.1 Philips Clear Vue 550 198
8.11.6.2 GE Logiq P5 199
8.11.6.3 Samsung H60 199
8.11.6.4 Alpinion Ecube 7 199
8.11.7 Portable Systems 200
8.11.7.1 GE Voluson 1 200
8.11.7.2 Samsung UGEO HM70A 200
8.12 Therapeutic Genome Editing: A Breakthrough Technology 201
8.12.1 Genome Editing Technologies 201
8.12.1.1 Growing Popularity of CRISPR Kits 201
8.12.2 Therapeutic Applications of Genome Editing 202
8.12.3 The First Genetically Modified Human Embryos 203
9.0 Prenatal Screening and Diagnostics: Market Overview 204
9.1 Global Market for Fetal Ultrasound 204
9.1.1 Market Leaders in Fetal Ultrasound 206
9.2 Global Market for Prenatal MRI Screening 207
9.3 Global Market for Maternal Serum Screening Tests 208
9.4 Global Market for Noninvasive Prenatal Tests (NIPTs) using cfDNA 210
9.4.1 Global Market for NIPTs by Product 211
9.5 Global Market for Prenatal Diagnostic Invasive Tests 212
9.6 Global Market for Newborn Screening for Genetic Diseases 213
9.6.1 Newborn Screening Market by Technology 214
9.7 Global Market for Preimplantation Screening/Diagnosis (PGS/PGD) 219
10.0 Other Genetic Testing-Related Markets 221
10.1 Emerging Trends in Molecular Diagnostics Market 221
10.2 Emerging Trends in Liquid Biopsy Market 222
10.3 Emerging Trends in Personalized Medicine Diagnostics Market 224
10.4 Emerging Trends in in vitro Diagnostics (IVD) Market 226
11.0 Market SWOT and Strategy Analysis 228
11.1 Acquisition Activities in Prenatal Screening Industry 228
11.1.1 Acquisition of BlueGenome by Illumina 228
11.1.2 Illumina’s Acquisition of Verinata 228
11.1.3 Eurofins Joint Venture with Emory Genetics Laboratory 228

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11.1.4 Roche’s Acquisition of Ariosa Diagnostics 228
11.1.5 Cooper Surgical’s Acquisition of Reprogenetics 228
11.2 Market Drivers 229
11.3 Market Restraints 229
11.4 Future of Prenatal Screening 229
11.5 Legislation Mandating Newborn Screening by Geography 229
11.5.1 Newborn Screening Programs in Americas 230
11.5.2 Newborn Screening Programs in Asia/Pacific 230
11.5.3 Newborn Screening Programs in Europe 230
11.5.4 Newborn Screening Programs in Middle East and Northern Africa 231
11.6 Ethical Voices against Problematic Applications of NIPT and NIPD 231
11.7 Clinical Implementation of NIPTs 232
11.7.1 Implementation of NIPTs in Developed Countries 232
11.7.3 Implementation of NIPTs in Developing and Under-Developed Countries 233
11.8 Adoption Rates for Different Prenatal Tests in the U.S. 233
12.0 Company Profiles 234
12.1 23andMe Inc. 234
12.1.1 23andMe’s Agreement with Pfizer 235
12.1.2 23andMe’s Agreement with Genentech 235
12.1.3 FDA’s Approval for 23andMe’s Screening Test for Bloom Syndrome 235
12.2 Abbott Laboratories 235
12.2.1 ARCHITECT AFP Assay 236
12.3 Abbott Molecular Inc. 236
12.3.1 AneuVysion 236
12.3.2 Cystic Fibrosis Genotyping Assay 236
12.4 Abcam plc 237
12.4.1 hCG Human ELISA Kit 237
12.5 AB Sciex LLC 237
12.6 Adaltis S.r.l 238
12.6.1 CLIAGen Free Beta-HCG Kit 238
12.7 Adaptive Biotechnologies Corp. 238
12.7.1 ImmunoSEQ Platform 238
12.7.2 ClonoSEQ 239
12.7.3 Pipeline 239
12.7.3.1 Quantifying Tumor Infiltration Lymphocytes in Solid Tumors 239
12.7.3.2 Measuring Immune Reconstitution Post Transplant 239
12.7.3.3 Diagnosing Cutaneous T-Cell Lymphoma (CTCL) 239
12.7.3.4 Therapeutics 239
12.8 Affymetrix Inc. 240
12.8.1 Microarray Solutions 240
12.8.2 Affymetrix’s Collaborating Partners 241
12.8.3 Affymetrix’s R&D Investment 241
12.9 Agena Biosciences Inc. 242
12.9.1 MassARRAY System 242
12.9.2 Acquisition of Sequenom’s Bioscience Business by Agena Biosciences 242
12.10 Agilent Technologies Inc. 243
12.10.1 SureScan Microarray Scanner 243
12.10.2 OneSeq 243
12.11 Ambyr Genetics Corp. 244
12.11.1 Clinical Diagnostic Tests 244
12.12 Analogic Corp. 245
12.12.1 Flex Focus 500 245
12.12.1 SonixTouchQ+ 246
12.12.3 SonixSP Q+ 246
12.13 Ariosa Diagnostics Inc. 246

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12.13.1 Harmony Test 246
12.13.2 Granting of Licenses to Harmony Test 247
12.14 Ansh Labs 247
12.14.1 picoPAPP-A ELISA Kit 247
12.14.2 Inhibin A ELISA Kit 247
12.15 Appistry Inc. 248
12.15.1 GenomePilot 248
11.15.2 Ayrris for NGS Analysis 248
12.15.3 Variant Annotation and Analysis Suite 248
12.15.4 CloudDx Translational 248
12.15.5 CloudDx Clinical 248
12.15.6 Appistry’s Partnership with LifeMap Sciences 249
12.16 ARUP Laboratories 249
12.16.1 Prenatal Screening and Diagnosis 249
12.16.2 Next-Generation Sequencing (NGS) 249
12.16.3 Noninvasive Prenatal Test for Aneuploids 250
12.17 AssureRx Health Inc. 250
12.17.1 GeneSight Psychotropic 250
12.17.2 GeneSight Analgesic 250
12.17.3 GeneSight ADHD 250
12.17.4 GeneSight MTHFR 251
12.18 Asuragen Inc. 251
12.18.1 AmplideX FMR1 PCR 251
12.18.2 AmplideX FMR1 mPCR 251
12.18.3 AmplideX Controls 251
12.18.4 Quantidex Pan Cancer Kit 251
12.18.5 Quantidex BCR/ABL1 Quant Kit 252
12.18.6 Quantidex DNA Assay 252
12.18.7 Signature Technology 252
12.18.8 Signature KRAS Mutations 252
12.18.9 Signature BRAF Mutations 252
12.18.10 Signature LTx v2.0 252
12.18.11 Signature NPM1 Mutations 253
12.19 Athena Diagnostics Inc. 253
12.20 AutoGenomics Inc. 253
12.20.1 INFINITI System 254
12.21 Base4 Innovation Ltd. 254
12.21.1 Microdroplet Sequencing 254
12.22 Beckman Coulter Inc. 254
12.22.1 SPRiworks System I for Illumina Genome Analyzer 255
12.22.2 SPRiworks HT for Illumina NGS Platform 255
12.22.3 SPRiselect Reagent Kit 255
12.22.4 Automated Sample Preparation 255
12.22.5 Genomic Services 255
12.22.6 Access Total Beta-hCG Reagent 255
12.22.7 Access Inhibin A Assay 255
12.23 Berry Genomics Co., Ltd. 256
12.23.1 Bambni Test 256
12.23.2 Berry’s Partnership with Illumina 256
12.24 Bina Technologies Inc. 256
12.24.1 Bina RAVE 257
12.24.2 Bina AAiM 257
12.24.3 Bina’s Collaboration with AsraZeneca 257
12.25 Bio-Rad (Israel) Laboratories Inc. 257
12.26 BGI 258
12.27 Blueprint Genetics Oy 259
12.27.1 Services 259
12.28 Boreal Genomics Inc. 259
12.28.1 OnTarget Mutation Detection System 259
12.28.2 Aurora Platform 260
12.29 Cambridge Epigenetics Ltd. 260
12.29.1 TrueMethyl Seq Kit 260
12.29.2 TrueMethyl Array 260
12.30 Caris Life Sciences 261
12.30.1 ADAPT Biotargeting System 261
12.30.2 Caris Molecular Intelligence 261
12.30.3 Collaboration between Caris Life Sciences and Syapse 261
12.31 CeGaT GmbH 262
12.31.1 NGS Service 262
12.31.2 ADME Research Panel 262
12.31.3 Forschungsexom 262
12.32 Centogene AG 263
12.33 Chromsystems Instruments & Chemicals GmbH 263
12.34 Chronix Biomedical Inc. 264
12.34.1 Technology 264
12.35 Claritas Genomics Inc. 264
12.36 CLC bio A/S 267
12.36.1 Products 267
12.37 Combimatrix Corp. 268
12.37.1 CombiSNP Array for Prenatal Diagnosis 268
12.37.2 CombiPGS 268
12.38 Contec Medical Systems Co., Ltd. 274
12.38.1 CMS600P B-Ultrasound Diagnostic System 274
12.38.2 CMS600B3 B-Ultrasound Diagnostic System 274
12.39 Coriell Life Sciences Inc. 274
12.39.1 GeneDose 274
12.40 Correlagen Diagnostics Inc. 275
12.41 Counsyl Inc. 277
12.41.1 Family Prep Screen 277
12.41.2 Informed Pregnancy Screen 277
12.41.3 Inherited Cancer Screen 277
12.42 Courtagen Life Sciences Inc. 278
12.43 Creative Diagnostics 278
12.43.1 Alpha-Fetoprotein, AFP ELISA Kit 278
12.43.2 hCG ELISA Kit 279
12.43.3 Human Free Estriol ELISA Kit 279
12.44 Cynvenio Biosystems Inc. 279
12.44.1 LiquidBiopsy 279
12.44.2 ClearID 279
12.44.3 Thermo Fisher’s Distribution Agreement with Cynvenio 280
12.44.4 Cynvenio’s Collaboration with the University of Southern Denmark 280
12.45 Diagnostic Automation/Cortez Diagnostics Inc. 280
12.46 Demeditec Diagnostics GmbH 281
12.46.1 Estriol, Free/Uncoujugated ELISA 281
12.47 DRG International Inc. 281
12.47.1 DRG PAPP-A ELISA 281
12.48 DNA Electronics Ltd. 282
12.48.1 Genalysis 282
12.49 DNA Link Inc. 282
12.49.1 Services 282
12.49.2 Partnership between Affymetrix and DNA Link Inc. 283
12.49.3 DNAGPS 283
12.49.4 AccuID 283
12.50 DNAnexus Inc. 284
12.50.1 Adoption of DNAnexus’ Cloud Genomics Platform by Natera 284
12.51 Eagle Biosciences Inc. 284
12.51.1 Alport Syndrome Kit 285
12.51.2 Free Beta-hCG ELISA 285
12.52 Eagle Genomics Ltd. 285
12.52.1 EagleCore 285
12.52.2 EagleNsembl 285
12.53 Enzymatics Inc. 286
12.53.1 Reagents 286
12.53.2 SPARK DNA Sample Preparation Kit 286
12.54 Esoate SpA 286
12.54.1 MyLabGamma 287
12.54.2 MyLab GOLD Platform 287
12.54.3 MyLab 50 287
12.54.4 MyLab 40 287
12.54.5 MyLab 25Gold 287
12.55 Eurofins MWG Operon Inc. 287
12.55.1 Products and Services 288
12.56 Exiqon A/S 289
12.56.1 Products and Services 289
12.57 Fakuda Denshi Co., Ltd. 290
12.57.1 UF-400AX 290
12.57.2 UF-550XTD 290
12.57.3 UF-760AG 291
12.57.4 UF-870AG 291
12.58 GATC Biotech AG 292
12.58.1 Sanger Sequencing 292
12.59 GE Healthcare Ltd. 292
12.59.1 VScan 293
12.59.2 VScan with Dual Probe 293
12.59.3 Logiq P5 293
12.59.4 Logiq P6 293
12.59.5 GE Venue 50 293
12.59.6 GE Venue 40 293
12.59.7 Logiq S8 293
12.59.8 Logiq E9 294
12.60 GenapSys Inc. 294
12.60.1 GENIUS 294
12.61 Gene by Gene Ltd. 294
12.62 Genecision Inc. 295
12.63 GeneDx Inc. 296
12.63.1 Cytogenetics and Biochemical Tests 296
12.63.2 Deletion/Duplication Analysis 296
12.63.3 Carrier Mutation-Specific Testing 297
12.64 GenePeeks Inc. 297
12.64.1 Matchright Technology 297
12.65 Genesis Genetics 298
12.65.1 GeniSeq24 298
12.65.2 UltraPGD 298
12.65.3 ArrayCGH 298
12.66 Genetadi Biotech S.L. 299
GLOBAL PRENATAL & MATERNAL DIAGNOSTIC MARKET TO 2020

12.66.1 AMNIOCHIP 299
12.66.2 FERTICHIP 299
12.66.3 PRENATAL GENE 299
12.67 Genoma Group Srl 300
12.68 Genomed AG 300
12.68.1 Aneufast 300
12.69 GenPath Diagnostics 301
12.69.1 InheriGen, InheriGen Plus and InheriGen Tx 301
12.69.2 Prenatal Tests 301
12.70 Genway Biotech Inc. 301
12.71 Good Start Genetics Inc. 302
12.71.1 EmbryVu 302
12.72 Hitachi Medical Systems America Inc. 302
12.72.1 Arietta 70 302
12.72.2 ProSound Alpha 7 302
12.72.3 Noblus 303
12.72.4 ProSound F37 303
12.73 Hologic Inc. 303
12.73.1 InPlex CF Molecular Test 303
12.73.2 Rapid fFN Test 303
12.74 Illumina Inc. 304
12.74.1 The Verifi Prenatal Test 304
12.75 INEX Innovations Exchange Pte Ltd. 305
12.75.1 iGene 306
12.75.2 FlashFISH 306
12.75.3 INEX’s Collaborative Agreement with BGI 306
12.76 Invitae Corp. 306
12.77 Laboratory Corporation of America Inc. 307
12.77.1 Services 307
12.78 LifeCodexx AG 308
12.78.1 Prena Test 308
12.79 Monobind Inc. 308
12.79.1 Fertility VAST Panel: HCG, FSH, LH, SPRL 308
12.79.2 PAPP-A Test 309
12.80 Multiplicom NV 309
12.80.1 Clarigo309
12.81 Myriad Genetics 309
12.82 Natera Inc. 310
12.82.1 Panorama Prenatal Screen 310
12.82.1.1 Accuracy of Panorama Test 310
12.82.1.2 Minimal False Negatives in Panorama 311
12.82.2 Horizon Carrier Screening 311
12.82.3 Spectrum Preimplantation Test 312
12.82.4 Natera’s 24-chromosome PGS 313
12.82.5 Anora Miscarriage Test 313
12.82.6 Prenatal Paternity Test 313
12.82.7 Agreement between Natera and LifeLabs 313
12.83 Natus Medical Inc. 314
12.83.1 ABAer 314
12.83.2 Algo 5 314
12.83.3 Algo 3i 314
12.83.4 AuDX 315
12.83.5 Echo-Screen III 315
12.84 NewGene Ltd. 315
12.84.1 Hereditary Disorders 316

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12.84.2 Alport Syndrome 316
12.84.3 Aortopathy Panel 316
12.84.4 RASopathy Gene Panel 316
12.84.5 Familial Hypercholesterolaemia 317
12.85 NIPD Genetics Ltd. 317
12.85.1 VERAILITY Test (NIPT) 317
12.86 Oxford Gene Technology (OGT) 318
12.86.1 SureSeq NGS Library Preparation Kit 318
12.86.2 CytoSure Embryo Screen Array 318
12.86.3 Cytocell FISH Probes 318
12.86.4 CytoSure Embryo Screen Array 318
12.86.5 CytoSure Aneuploidy Array 318
12.86.6 CytoSure Chromosome X Arrays 319
12.87 Parabase Genomics Inc. 319
12.87.1 NewbornDx Test 319
12.88 Pathway Genomics Corp. 320
12.88.1 Carrier Screening 320
12.89 PerkinElmer Life and Analytical Sciences Inc. 320
12.89.1 DELFIA Xpress PIGF Assay 320
12.89.2 DELFIA Xpress PAPP-A Kit 320
12.89.3 AutoDELFIA PAPP-A Kit 321
12.89.4 StepOne Newborn Screens 321
12.90 Premaitha Health PLC 321
12.90.1 IONA Test 321
12.90.2 Premaitha’s Investment Agreement with Thermo Fisher 322
12.91 Progenity Inc. 322
12.91.1 nXtPanel Test 322
12.91.2 Verifi Test 322
12.91.3 Acquisition of Carmenta Bioscience by Progenity 323
12.92 Qiagen N.V. 323
12.92.1 Next-Generation Sequencing (NGS) 323
12.92.2 Target Enrichment Solutions 324
12.92.3 Library Construction 324
12.93 Quest Diagnostics Inc. 324
12.93.1 QNatal Advanced 324
12.93.2 Inhibin A 325
12.94 Ravgen Inc. 325
12.94.1 Prenatal Downs Syndrome Testing 325
12.94.2 Single Gene Disorder Testing 325
12.95 Recombine Inc. 325
12.95.1 CarrierMap 326
12.96 Reproductive Genetics Institute Inc. 326
12.97 Reprogenetics Laboratories 326
12.97.1 Array CGH (aCGH) 326
12.97.2 Single Gene Disorder Testing 326
12.97.3 WaferGen Biosystems’ Supply Agreement with Reprogenetics 327
12.98 Sebia Inc. 327
12.99 Sequenom Inc. 327
12.99.1 MaterniT GENOME Test 328
12.99.1.1 Sensitivity and Specificity of MaterniT GENOME Test 328
12.99.2 MaterniT21 PLUS 329
12.99.2.1 Sensitivity and Specificity of MaterniT21 PLUS 329
12.99.3 Heredit UNIVERSAL Carrier Screen 330
12.99.4 VisibiliT Prenatal Test 330
12.99.5 Heredit Cystic Fibrosis Carrier Screen 330
12.99.5.1 Clinical Data for HerediT 331
12.99.6 SensiGene Fetal RHD Genotyping 331
12.99.7 NextView Prenatal Diagnostic Tests 331
12.100 SeraCare Life Sciences Inc. 332
12.100.1 Seraseq Trisomy 21 Aneuploidy Reference Material 332
12.101 Siemens Healthcare 333
12.101.1 Acuson S2000 333
12.101.2 Acuson X700 333
11.101.3 Acuson X600 333
12.101.4 Acuson X300 PE 333
12.101.5 Acuson X150 334
12.101.6 Acuson Antares 334
12.101.7 Acuson P300 334
12.102 Sophia Genetics SA 334
12.102.1 Clinical Genomic Modules 334
12.103 SpOtOn Clinical Diagnostics Ltd. 335
12.103.1 Newborn Screening 335
12.103.2 Ante-Natal Testing 335
12.104 Stra Biotech GmbH 335
12.105 Sygnis AG 336
12.105.1 TruePrime Single Cell WGA Kit 336
12.105.2 TruePrime WGA Kit 336
12.105.3 TruePrime RCA Kit 336
12.105.4 SunScript Reverse Transcriptase RNaseH+ and RNaseH- Kits 336
12.105.5 SensiPhi/QualiPhi 336
12.106 SynapDx Corporation 337
12.106.1 ASD Diagnosis 337
12.107.1 Thermo Fisher’s Brands 337
12.108 Transgenomic Inc. 338
12.108.1 Technology Suite 338
12.108.2 MX-ICP Technology 338
12.108.3 ICEme Kit 339
12.108.4 Other Products 339
12.109 Trivitron Healthcare Private Ltd. 339
12.110 Trovagene Inc. 340
12.110.1 PCM BRAF V600E Mutation Detection Test 340
12.110.2 PCM KRAS Mutation Detection Test 340
12.110.3 PCR EGFR Mutation Detection Test 340
12.110.4 HPV HR Detection Test 341
12.111 Tute Genomics Inc. 341
12.111.1 Tute Platform 342
12.112 Warp Drive Bio LLC 342
12.112.1 Wrap’s New Approach 342
12.112.2 Chemomemes 342
12.112.3 Genomic Search Engine 342
12.113 ZS Genetics Inc. 343
12.113.1 3G Sequencing 343
12.114 Zymo Research Corporation 343
12.114.1 DNA Methylation Products 343
12.114.1.1 Bisulfite Conversion 343
12.114.1.2 Methylated DNA Standards 344
12.114.1.3 DNA Methyltransferases 344
12.114.1.4 5-mC ELISA 344
12.114.1.5 5-mC Antibodies and Immunoprecipitation 344
11.114.1.6 Region-Specific DNA Methylation Analysis 344
GLOBAL PRENATAL & MATERNAL DIAGNOSTIC MARKET TO 2020

12.114.1.7 Global 5-mC Quantification 344
12.114.1.8 Genome-wide 5-mC Analysis 345
12.114.2 DNA Purification Products 345
12.114.2.1 DNA Clean-up 345
12.114.2.2 Plasmid DNA Purification 345
12.114.2.3 Genomic DNA 345
12.114.2.4 Microbial and Environmental DNA Isolation 346
12.114.2.5 DNA/RNA Co-Purification 346
12.114.2.6 Sample Collection and Stabilization 346
12.114.2.7 DNA Ladders 346
12.114.2.8 Enzymes 346
12.114.2.9 High-Throughput/Automated Isolation 347
12.114.2.10 DNA Analysis Kits 347
12.114.3 Other Products 347

APPENDIX

Appendix 1: Representative Companies in Prenatal and Maternal Diagnostics and their Products 348
Appendix 2: Sources for Tables and Graphs 351

INDEX OF FIGURES

Figure 1.1: Summary of Prenatal Screening, Prenatal Diagnosis, Newborn Screening and PGS/PGD Markets, Through 2021 24
Figure 2.1: Risk of Down Syndrome and Chromosomal Abnormalities Due to AMA 28
Figure 2.2: Maternal Age and Pregnancy Loss Rate 29
Figure 2.3: Risk of Cesarean Section with Increasing AMA 30
Figure 2.4: Risk of Gestational Diabetes with AMA 31
Figure 2.5: Risk of Placenta Previa with AMA 32
Figure 2.6: Delayed First Pregnancy in the U.S. 33
Figure 2.7: First Birth Rates for 35-39 Aged U.S. Women by Race 34
Figure 2.8: Pre-Existing Chronic Diseases among Women of Reproductive Ages in the U.S. 41
Figure 2.9: Chronic Disease Risk Behaviors and Risk Factors among Women of Reproductive Ages in the U.S. 41
Figure 2.10: Prevalence of Preeclampsia during Pregnancy by Race/Ethnicity in the U.S. 43
Figure 2.11: Prevalence of Preeclampsia by Maternal Age in the U.S. 43
Figure 2.12: Prevalence of Gestational Diabetes by Maternal Age in the U.S. 45
Figure 2.13: Prevalence of Gestational Diabetes by Maternal Race/Ethnicity in the U.S. 45
Figure 2.14: Causes of Preterm Labor 46
Figure 2.15: Prevalence of Preterm Births in the U.S. by Stage 48
Figure 2.16: Prevalence of Preterm Births in the U.S. by Race/Ethnicity 49
Figure 2.17: Prevalence of Preterm Births in the U.S. by Plurality of Birth 50
Figure 2.18: Prevalence of Preterm Births in the U.S. by Maternal Age 51
Figure 2.19: Prevalence of Multiple Births in the U.S. 51
Figure 3.1: Inheritance of Autosomal Dominant Faulty Gene When One Parent is a Carrier 59
Figure 3.2: Inheritance of Autosomal Dominant Faulty Gene When Both Parents are Carriers 60
Figure 3.3: Percent of Patients taking Sweat Chloride Test Reported in the U.S. Registry 61
Figure 3.4: Age at CF Diagnosis for all People Reported in the U.S. Registry, 2013 62
Figure 3.5: Distribution of Race/Ethnicity among People with CS in the U.S., 2013 62
Figure 3.6: Percent of New CF Diagnosis in U.S. Newborn Screenings, 1990-2013 63
Figure 3.7: The Six Most Common CFTR Mutations in the U.S. 64
Figure 3.8: Autosomal Recessive Inheritance when Both Parents are Carriers 71
Figure 3.9: Autosomal Recessive Inheritance with Carrier Mother and Non-Carrier Father 71
Figure 3.10: Autosomal Recessive Inheritance from Homozygous Recessive Mother and Carrier Father 72
GLOBAL PRENATAL & MATERNAL DIAGNOSTIC MARKET TO 2020

Figure 9.18: Global Market for PGS/PGD by Geography (North America, Europe, RoW) 220
Figure 10.1: Global Market for Molecular Diagnostics, Through 2021 222
Figure 10.2: Global Market for Liquid Biopsy by Geography (North America, Europe, RoW), Through 2021 224
Table 10.3: Global Market for Personalized Medicine by Business Segment, Through 2021 225
Figure 10.4: Percent Share of IVD Market by Business Segments 226
Figure 10.5: Global IVD Market by Geography (North America, Europe, RoW), Through 2021 227
Figure 12.1: Comparison of False Negative Rates 311
Figure 12.2: Sensitivity and Specificity of MaterniT GENOME Test 328

INDEX OF TABLES

Table 1.1: Summary of Prenatal Screening, Prenatal Diagnosis, Newborn Screening and PGS/PGD Markets, Through 2021 28
Table 2.1: Risk of Down Syndrome and Chromosomal Abnormalities Due to AMA 29
Table 2.2: Maternal Age and Pregnancy Loss Rate 30
Table 2.3: Risk of Cesarean Section with Increasing AMA 31
Table 2.4: Risk of Gestational Diabetes with AMA 32
Table 2.5: Risk of Placenta Previa with AMA 33
Table 2.6: Delayed Fist Pregnancy in the U.S. 34
Table 2.7: First Birth Rates for 35-39 Aged U.S. Women by Race 35
Table 2.8: Maternal and Fetal Concerns due to Pre-Existing Maternal Medical Issues 36
Table 2.9: Pregnancy Outcome in Women with Renal Diseases 37
Table 2.10: Pregnancy Outcome in Women with Pre-Existing Thyroid Disease 38
Table 2.11: Risks Associated with Obesity in Pregnant Women 39
Table 2.12: Pregnancy Complications in Asthmatic Women 40
Table 2.13: Pregnancy Complications and Neonatal Outcome in Women with Epilepsy 41
Table 2.14: Maternal and Fetal Complications in Women with Autoimmune Diseases 42
Table 2.15: Complications due to Hemoglobinopathies in Pregnancy 43
Table 2.16: Prevalence of Gestational Diabetes in the U.S. 44
Table 2.17: Countries with Higher and Lower Rates of Preterm Births 45
Table 2.18: Prevalence of Preterm Births in the U.S. by Stage 46
Table 2.19: Prevalence of Preterm Births in the U.S. by Race/Ethnicity 47
Table 2.20: Prevalence of Preterm Births in the U.S. by Plurality of Birth 48
Table 2.21: Prevalence of Preterm Births in the U.S. by Maternal Age 49
Table 2.22: Related Complications of Placenta Previa 50
Table 2.23: Mode of Delivery for Breech Presentation 51
Table 2.24: Perinatal Outcome due to Meconium Stained Liquor 52
Table 2.25: Pregnancy Outcome due to Polyhydramnios 53
Table 2.26: Causes of Fetal Growth Restriction 54
Table 2.27: Prevalence of Congenital Cardiovascular Diseases in the U.S. 55
Table 3.1: Worldwide Prevalence of Huntington Disease by Selected Country 56
Table 3.2: Prevalence of 25 Most Common CFTR Mutations in the U.S., 2013 57
Table 3.3: Molecular Diagnostic Tests for Canavan Disease 58
Table 3.4: Incidence of Sickle Cell Disease in the U.S. 59
Table 3.5: Red Blood Cell Indices in Beta-Thalassaemia 60
Table 3.6: Molecular Genetic Tests for Beta-Thalassaemia 61
Table 3.7: Chances of Inheriting a Single Gene Disorder 62
Table 3.8: More Common Recessive Disease Traits in Selected Ethnic Groups 63
Table 3.9: A Comprehensive List of Genetic Disorders 64
Table 3.10: Turnaround Time and Average Cost for Hemophilia Genetic Tests 65
Table 3.11: Incidence of Chromosomal Abnormalities 66
Table 3.12: Karyotypes Commonly Associated with Klinefelter Syndrome 67
Table 3.13: Features of Turner Syndrome in Different Age Groups 68
Table 3.14: Some Complications of Trisomy 21 (Down Syndrome) 69
Table 3.15: Turnaround Time and Average Cost for Hemophilia Genetic Tests 70
Table 3.16: Prevalence of Congenital Cardiovascular Diseases in the U.S. 71
Table 3.17: Worldwide Prevalence of Huntington Disease by Selected Country 72
Table 3.18: Prevalence of 25 Most Common CFTR Mutations in the U.S., 2013 73
Table 3.19: Molecular Diagnostic Tests for Canavan Disease 74
Table 3.20: Incidence of Sickle Cell Disease in the U.S. 75
Table 3.21: Red Blood Cell Indices in Beta-Thalassaemia 76
Table 3.22: Molecular Genetic Tests for Beta-Thalassaemia 77
Table 3.23: Chances of Inheriting a Single Gene Disorder 78
Table 3.24: More Common Recessive Disease Traits in Selected Ethnic Groups 79
Table 3.25: A Comprehensive List of Genetic Disorders 80
Table 3.26: Turnaround Time and Average Cost for Hemophilia Genetic Tests 81
Table 3.27: Incidence of Chromosomal Abnormalities 82
Table 3.28: Karyotypes Commonly Associated with Klinefelter Syndrome 83
Table 3.29: Features of Turner Syndrome in Different Age Groups 84
Table 3.30: Some Complications of Trisomy 21 (Down Syndrome) 85

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Table 3.15: Incidence of Down Syndrome by Maternal Age
Table 3.16: Developmental Delay in Children with Down Syndrome
Table 3.17: Frequently Observed Anomalies in Babies with Trisomy 18
Table 3.18: Common Clinical Features of Trisomy 13
Table 3.19: Congenital Anomalies Associated with Triploidy
Table 3.20: Relative Frequencies of Different Chromosomal Anomalies in Spontaneous Abortions
Table 3.21: Maternal Age-Related Frequencies of Aneuploid Fetuses Detected Prenatally
Table 3.22: Clinical Features of Common Chromosomal Aneuploidy
Table 3.23: Maternal Age and Chromosomal Aneuploidy
Table 3.24: Sensitivity and Specificity of Maternal Serum and Ultrasound Tests for Aneuploids
Table 3.25: Common Autosomal Deletions, Resulting Syndromes and Clinical Features
Table 3.26: Common Autosomal Microdeletion Syndromes
Table 3.27: Commonly Encountered Problems in Prader-Willi Syndrome
Table 3.28: Tests for Prader-Willi Syndrome
Table 3.29: Molecular Genetic Tests for CATCH 22 Syndrome
Table 3.30: Autosomal Duplication Syndromes
Table 3.31: Molecular Genetic Tests for Beckwith-Wiedemann Syndrome
Table 3.32: Molecular Diagnostic Tests for CMT1A
Table 4.1: Genetic Counseling Services by DTC GT Companies
Table 4.2: A List of Genetic Counseling Companies and Their Websites
Table 5.1: Major Prenatal Screening Tests during the First and Second Trimesters
Table 5.2: Other Routine and Non-Routine Prenatal Screening Tests
Table 5.3: hCG Levels from Date of Conception to 40th Week
Table 5.4: Selected β-hCG Kits and Manufacturers
Table 5.5: Selected PAPP-A Kits and Manufacturers
Table 5.6: Selected MSAFP Test Kits and Manufacturers
Table 5.7: Selected uE3 Test Kits and Manufacturers
Table 5.8: Selected Inhibin-A Test Kits and Manufacturers
Table 5.9: Selected Hexosaminidase Test Kits and Manufacturers
Table 5.10: Comparison of the Four Maternal Serum Screening Analyte Patterns
Table 5.11: The Five Popular Commercially Available Noninvasive Prenatal Tests (NIPTs)
Table 5.12: Sensitivity and Specificity of NIPTs
Table 5.13: Diagnostic Applications of cfDNA
Table 5.14: Time of Availability of cfDNA in Maternal Blood
Table 5.15: The Costs of NIPTs by Region/Country
Table 5.16: Costs of NIPTs by Product in the U.S.
Table 5.17: History of Aneuploidy Screening
Table 5.18: NIPT Methods for Detecting Aneuploidy
Table 5.19: Comparison of Amniocentesis and NIPTs
Table 5.20: Advantages and Disadvantages of NIPTs
Table 5.21: Comparison of Performance Criteria for Commonly Used Prenatal Tests
Table 5.22: Comparison of NIPT Detection Rates with Traditional Tests
Table 5.23: False Positive Rates of NIPTs Compared with those of Traditional Tests
Table 5.24: Different Applications of Ultrasound in Different Trimesters
Table 5.25: MRI Indications for Prenatal Screening when Ultrasound Screening is Inadequate
Table 5.26: Prenatal Diagnostic Tests by Pregnancy Stage
Table 5.27: Comparison of CVS and Amniocentesis
Table 5.28: Advantages and Disadvantages of Prenatal Diagnostic Tests
Table 5.29: Comparison of Prenatal Tests using Cell-Free Fetal DNA (cfDNA) and Intact Fetal Cells from Maternal Blood
Table 6.1: Thirty One Core Conditions Detected During Newborn Screening
Table 6.2: Categories of Newborn Genetic Disorders
Table 6.3: Metabolic Genetic Disorders Detectable by MS/MS and their Scores
Table 6.4: Incidence of PKU by Region
Table 6.5: Molecular Genetic Tests for Biotidinase Deficiency
Table 7.1: Advantages and Disadvantages of Currently Available PGS/PGD Technologies 154
Table 7.2: Genetic Diseases Detected During PGS/PGD 154
Table 7.3: Currently Available Products for PGS/PGD 159
Table 7.4: Cost of in vitro Fertilization and Related Procedures 160
Table 8.1: Karyotype vs. Microarray 167
Table 8.2: The Format of FISH Test Result 170
Table 8.3: Some Commonly Used FISH-Based Tests 170
Table 8.4: Microdeletions/Microduplications Detectable by FISH 171
Table 8.5: FISH Probes and Functions 172
Table 8.6: Presentation Format of Array-CGH Test Report 173
Table 8.7: Harmony vs. Traditional Down Syndrome Tests 174
Table 8.8: Predictive Value, Sensitivity and Specificity of InformaSeq Test 176
Table 8.9: Detection Rates of Panorama Test 177
Table 8.10: NIFTY Test Options 178
Table 8.11: Clinical Data for IONA Test 179
Table 8.12: Sensitivity and Specificity of Verifi Test 180
Table 8.13: Sensitivity and Specificity of Verifi’s Microdeletion Panel 181
Table 8.14: Comparison of MaterniT GENOME Test and Karyotype 181
Table 8.15: Sensitivity and Specificity of MaterniT GENOME Test 182
Table 8.16: MaterniT21 PLUS’ Independent Validation 182
Table 8.17: The Three Panels Offered by HerediT UNIVERSAL Carrier Screen 183
Table 8.18: Comparison of the Four Major NIPT Products available in the U.S. Market 183
Table 8.19: U.S. Patents for NIPTs by Company 184
Table 8.20: Selected Issued Patents of NIPTs 185
Table 8.21: Major U.S. Patents and Applications 186
Table 8.22: Countries in Which NIPT is currently Marketed 187
Table 8.23: Detection, Uptake, False Positive and Failure Rates for NIPTs 188
Table 8.24: Cost of Different Prenatal Screens 188
Table 8.25: Disease Types Targeted by Different Nuclease Platforms 203
Table 9.1: Global Market for Fetal Ultrasound Screening by Geography (U.S., Europe, RoW), Through 2021 205
Table 9.2: Global Fetal Ultrasound Market Share by Company, 2014 206
Table 9.3: Global Market for MRI Prenatal Screening by Geography (U.S., Europe and RoW), Through 2021 208
Table 9.4: Global Market for Maternal Serum Screening Tests by Geography (U.S., Europe, RoW), Through 2021 209
Table 9.5: Global Market for NIPTs by Geography (North America, Europe, RoW), Through 2021 211
Table 9.6: Global Market for NIPTs by Product 211
Table 9.7: Global Market for Prenatal Invasive Diagnostic Tests by Geography (U.S., Europe, RoW), Through 2021 212
Table 9.8: Global Market for Newborn Screening by Technology, Through 2021 215
Table 9.9: Global Market for PGS/PGD by Geography (North America, Europe, RoW), Through 2021 223
Table 10.1: Global Market for NIPTs by Geography (North America, Europe, RoW), Through 2021 225
Table 10.2: Global Market for Personalized Medicine by Business Segment, Through 2021 226
Table 10.3: Global IVD Market by Geography (North America, Europe, RoW), Through 2021 226
Table 11.1: Adoption Rates for Different Prenatal Tests in the U.S. 233
Table 12.1: Financial Data for Affymetrix 241
Table 12.2: Selected Financial Data for Agilent Technologies 243
Table 12.3: Comparison of Harmony and Other Traditional Tests 247
Table 12.4: CombiSNP Prenatal Targeted Array Disorder List 268
Table 12.5: Genetic Tests Offered by Correlagen by Gene and Disease 275
Table 12.6: GeneDx’s Cytogenetics and Biochemical Tests 296
Table 12.7: Hologic’s Cytogenetic Data 304
Table 12.8: Performance of Verifi Test 304
1.0 Introduction

Prenatal tests (fetus, embryos and newborns) currently on the market analyze chromosomes, DNA, RNA, genes, and/or gene products to determine whether an abnormality is present that is causative of a specific disease. Since 2011, the launch of noninvasive prenatal tests (NIPTs) is revolutionizing the prenatal screening industry providing detection rates of >99% and false positive rates of <0.1%. These screening results are only indicative of the risk and not confirmative. To confirm positive results of NIPT screening tests, prenatal diagnostic tests such as amniocentesis and chorionic villus sampling (CVS) are performed. Prior to NIPTs, ultrasound and maternal serum tests were routinely used to screen fetusus for genetic abnormalities.

Globally, 50 countries have effective newborn genetic test programs, the implementation of which is in various stages. In the U.S., four million newborns are tested annually, with 3,000 found to have metabolic and genetic diseases. The purpose of newborn screening is to identify affected newborns quickly and provide them with treatment to prevent mental retardation, prolonged illness and death. The newborn genetic screening market is crowded with different technologies, the common being tandem mass spectrometry, pulse oximetry, enzyme-based assays, DNA assays and electrophoresis.

Preimplantation genetic screening and preimplantation genetic diagnosis (PGS/PGD) are genetic tests performed in eight-celled embryos before implantation during in vitro fertilization (IVF) for the detection of genetic abnormalities and sex. These tests can detect and diagnose chromosomal rearrangements, X-linked diseases and help in reducing the incidence of spontaneous abortions, increase implantation rates, prevent trisomic offsprings and avoid the risk of transmitting single gene disorders. However, PGS and PGD results are not 100% accurate and after pregnancy, if required the diagnostic tests such as amniocentesis and chorionic villus sampling (CVS) are performed. Prior to NIPTs, ultrasound and maternal serum tests were routinely used to screen fetusus for genetic abnormalities.

The most important development in the field of prenatal screening, is the introduction of NIPTs using cell-free fetal DNA (cffDNA) in 2011. Obstetricians and patients who are deterred by the 10% to 15% false positive rates of ultrasound screening and 5% false positive rates of maternal serum tests are eagerly embracing NIPTs which claim to have only <0.1% of false positive results.

Within just four years, molecular genetics firms have successfully produced eight different types of kits for the detection of genetic abnormalities in the fetus. These products have been gradually marginalizing the maternal serum tests and in another decade, maternal serum tests are expected to become obsolete. If, companies can develop NIPTs with a 100% accuracy rate, maternal serum tests,
amniocentesis and CVS will all be completely overshadowed by NIPTs. Despite the growing popularity of NIPTs, maternal serum tests still continue to have some market due to their low price. While the maternal serum tests are available for just $x to $x, NIPTs are priced between $x and $x. However, Sequenom reportedly sold over x tests in 2013 and so pricing does not seem to be a deterrent factor. The introduction of technologies such as digital PCR and next-generation sequencing (NGS) or massively parallel sequencing (MPS) have also enabled accurate estimation of very small differences in chromosome-specific sequences in maternal blood.

1.1 Executive Summary

According to WHO, nearly 140 million babies are born every year, out of which 5 million die in the first month of life, mostly in developing countries. For example, in India, about 5% to 15% of sick newborns have a metabolic disorder. This emphasizes the need for newborn screening for preventing disability and death by early intervention, follow-up and counselling.

Globally, the use of maternal serum marker screening and ultrasound imaging for the detection of chromosome aneuploidies and other birth defects constitute a routine part of prenatal care in the first and/or second trimesters. Yet, both of these techniques have the disadvantages of high false positive rates, varying from 2% to 7%. If the results of these tests show that a fetus is at increased risk of aneuploidy, invasive approaches such as chorionic villus sampling (CVS) or amniocentesis are recommended for diagnosis.

Fetal Ultrasound Screening Market

Today, ultrasound is used on average five times per pregnancy before delivery. The fetal ultrasound screening market generated revenues of about $x million in 2014 with the potential to earn $x million in 2021, growing at a CAGR of x%. Fetal MRI prenatal screening is performed rarely, only when the ultrasound screening provides an ambiguous result and to detect suspected central nervous system (CNS) disorders in the fetus during the second trimester. The fetal MRI segment had generated estimated revenue of $x million in 2014 with a potential to earn $x million in 2021.

Maternal Serum Testing Market

The global maternal serum test market was worth $x million in 2014 and is forecast to grow and reach $x million by 2021. The market consists of test methods for:

- Pregnancy-associated plasma protein (PAPP-A)
- Alpha-fetoprotein (AFP)
- Human chorionic gonadotropin (hCG)
- Estriol
- Inhibin-A

Had it not been for the launch of noninvasive prenatal tests (NIPTs) in 2011, this market would have fared much better.

Noninvasive Prenatal Test Market

Noninvasive prenatal tests (NIPTs) are the future of prenatal screening and have shown signs of surpassing the traditional maternal serum test revenues by earning about $x million in 2014. With a CAGR of x%, this market is forecast to reach $x million in 2021.

Newborn Screening Market
Newborn screening is an accepted national health policy in about 52 countries. It is a well-entrenched routine of newborn care in the U.S., U.K., Japan, Australia and most western European countries for the last three decades. In the Asian continent, China, Philippines and Thailand have developed admirable newborn screening programs in a relatively short period of time. Mass spectrometry has become the standard technique for newborn screening. Using this technique, medical technologists are capable of screening as many as 30 metabolic disorders from a single sample of blood. The global market for newborn screening has been valued at $x million in 2014 and it will be worth about $x million by 2021.

**Preimplantation Genetic Diagnosis Market**

Preimplantation genetic diagnosis (PGD) has been in practice for more than twenty years in about 60 countries globally. PGD testing is performed to identify genetic defects in embryos. The tests are usually performed in cases of a known genetic defect, in late age pregnancy, or in patients with a history of repeated miscarriages. PCR, FISH, CGH, and SNP analysis are some of the most common techniques used, and PCR is the most widely used technology. The global market for PGD was worth about $x million in 2014 and this has been predicted to enlarge and reach $x million in 2021.

### 1.2 Objectives of this Report

Since the 1960s, technologies for testing fetuses for conditions including trisomies have been continually advancing. The initial nongenetic testing such as ultrasound and serum screening for protein level markers, have developed remarkably throughout the years. Yet, these conventional tests are limited in sensitivity and specificity. Parallel to the development of these conventional technologies, two invasive techniques called amniocentesis and chorionic villus sampling (CVS) were introduced in 1980s to offer near 100% detection rates for trisomies. Yet, the chances of injury to the fetus and the consequence of miscarriages deterred both patients and obstetricians from using these techniques and to this date the techniques are used in high-risk pregnancies for the confirmation of positive results from screening tests.

When human genome sequencing was successfully accomplished, experts in the healthcare industry expected that DNA sequencing would mainly focus on genetic diseases in adults. Instead, the impact of sequencing technology has been primarily on prenatal, newborn and PGD tests. In a short span of four years, the different types of maternal serum screening tests and fetal ultrasound tests are getting gradually replaced by the new technology of non-invasive prenatal tests (NIPTs). In the past four years, hundreds and thousands of these new tests have been performed in screening cffDNA from maternal blood.

The objective of this report is to provide an overview of the various technologies being employed for detecting genetic diseases in embryos, fetuses and newborns. The various chapters describe common pregnancy disorders, numerical chromosome disorders and structural chromosome disorders, single gene disorders inherited by fetuses, newborns and embryos. The global prenatal and maternal diagnostic test market is assessed with respect to:

- Fetal Ultrasound
- Prenatal MRI Screening
- Maternal Serum Screening Tests
- Noninvasive Prenatal Tests (NIPTs) using cffDNA
- NIPTs by Product
- Prenatal Diagnostic Invasive Tests
- Newborn Screening for Genetic Diseases
- Newborn Screening Market by Technology
- Preimplantation Screening/Diagnosis (PGS/PGD)
SWOT and merger/acquisition analysis is also performed as is a comprehensive documentation of the legislation pertaining to newborn screening by geography and how clinical programs are implemented in developed and developing markets.

Emerging trends in associated markets are also analysed in order to give the reader a comprehensive overview of how prenatal and maternal diagnostic testing is affected by the following industries:

- Molecular Diagnostics Market
- Liquid Biopsy Market
- Personalized Medicine Diagnostics Market
- In vitro Diagnostics (IVD) Market

This is a comprehensive 359 page report strengthened with over 260 figures and tables. Published in January 2016, the GLOBAL PRENATAL & MATERNAL DIAGNOSTIC MARKET TO 2020 report by KellySciPub has a detailed overview of 114 companies in the market with specifics pertaining to financial and business strategy, current products on the market and pipeline products.

1.3 Key Questions Answered in this Report

- What are the major prenatal pregnancy complications?
- What are the major genetic diseases detected in fetuses?
- What are the appropriate technologies for the detection of aneuploidies, microdeletions, duplications, copy number variations and translocations?
- How far is genetic counseling important in educating pregnant women and healthcare professionals?
- What is the impact of recent advances in clinical genomics on genetic counseling?
- What different noninvasive and invasive prenatal screening tests are performed during a pregnancy?
- What are the detection rate, true positive rate and true negative rate for NIPTs?
- Currently, in which countries are the NIPTs available?
- What is the cost of NIPTs region-wise?
- What is the uptake of conventional maternal serum tests, NIPTs and invasive diagnostic tests in the U.S.?
- What is the average cost of maternal serum screening, NIPTs, fetal ultrasound screening and invasive diagnostic tests in the U.S.?
- What are the strategies to be adopted for clinical implementation of NIPTs for all pregnancies?
- What is the “patient directed model” for the integration of NIPTs into healthcare systems?
- How does the detection rate of NIPTs compare with the rates of conventional maternal serum screening tests?
- What are the genetic disorders detectable by different prenatal screening and diagnostic tests?
- What is the reliability of amniocentesis and CVS results?
- Is the future of invasive diagnostic tests uncertain?
- Do the intact fetal cells in maternal blood have use in noninvasive prenatal diagnosis (NIPT)?
- What about the use of trophoblast cells obtained from cervix in NIPD?
- How many genetic conditions are detected during newborn genetic screening in the U.S.?
- What is the status of newborn genetic screening in developed, developing and other countries?
- What are the different preimplantation genetic screening (PGS) and preimplantation genetic diagnosis (PGD)?
- What are the currently available PGS/PGD testing products available in the market?
- What are the currently used advanced technologies in prenatal, newborn and PGD testing?
- What is the global market for fetal ultrasound screening, through 2021?
- Who are the market leaders in fetal ultrasound systems?
What is the global and regional market for prenatal screening by fetal MRI, through 2021?
What is the global and regional market for maternal screening tests, through 2021?
What is the global and regional market for NIPTs using cfDNA, through 2021?
What is the global and regional market for invasive prenatal diagnostic tests, through 2021?
What is the global and regional market for newborn screening tests, through 2021?
What is the global market for newborn screening tests by technology, through 2021?
What is the global and regional market for pre-implantation genetic diagnosis (PGD), through 2021?
What is the global market for molecular diagnostics, through 2021?
What is the global market for liquid biopsy tests, through 2021?
What is the overall global market for personalized medicine diagnostics, through 2021?
What is the overall global market for in vitro diagnostics (IVD), through 2021?
What are the recent acquisition activities in prenatal screening industry?
What are the important factors that drive the growth of prenatal & newborn genetic testing industry?
What are the challenges faced by the prenatal screening industry?
What are the possible future developments in prenatal screening industry?