

# ROLE OF THREE-DIMENSIONAL ULTRASONOGRAPHY IN PRENATAL DIAGNOSIS OF CRANIO-FACIAL ANOMALIES

*Essay*

*Submitted for Partial Fulfillment of  
Master Degree in Radiodiagnosis*

*By*

*Ayman Mohamed Mohamed Khalifa  
M.B., B.Ch.*

*Under supervision of:*

**Prof. Dr. Nivienne Mostafa Ibrahim**

*Professor of Radiodiagnosis*

*Faculty of Medicine -Ain Shams University*

**Prof. Dr. Sherine Kadry Amin**

*Professor of Radiodiagnosis*

*Faculty of Medicine -Ain Shams University*

**Ass. Prof. Dr. Ahmed Bassim Ali**

*Assistant Professor of Radiodiagnosis*

*Research Institute of Ophthalmology*



**2013**



# ACKNOWLEDGEMENT

Thanks to Allah for helping me in achieving this work and every other work in my life.

I wish to express my highest and deepest appreciation to: **Prof. Dr. Niviene Mostafa Ibrahim**, Professor of Radiodiagnosis, Faculty of Medicine, Ain Shams University, for her great efforts in supervising this work and for the valuable ideas, comments and advices. This thesis would not have been possible without her support and efforts.

I cannot find words to express my sincere gratitude to: **Prof. Dr. Sherine Kadry Amin**, Professor of Radiodiagnosis, Faculty of Medicine, Ain Shams University, for her persistent effort and patience, valuable guidance and meticulous revision of the work. I am really indebted and exceedingly grateful for her invaluable support to complete this work.

It is with immense gratitude that I acknowledge the support and help of **my Professor Dr. Ahmed Bassim Ali**, Assist. Professor of Radiodiagnosis, Research Institute of Ophthalmology. He always gives me the motivation and enthusiasm.

Last but not the least, I owe more than thanks to my parents and my family members, for their support and encouragement throughout my life.



# CONTENTS:

<b>I. Introduction &amp; aim of the work.....</b>	<b>1</b>
<b>II. Basic considerations:</b>	
Anatomy. Embryology. Pathogenesis.....	3
<b>III. Basics of 3D ultrasound technique.....</b>	<b>28</b>
<b>IV. Sonographic examination of the fetal face     and normal appearance. ....</b>	<b>45</b>
<b>V. Sonographic and clinical features of     craniofacial anomalies.....</b>	<b>70</b>
- Facial clefts .....	74
- Fetal eye anomalies .....	85
- Fetal ear anomalies.....	101
- Nasal bone, Facial angle &Down syndrome....	105
- Anomalies of the fetal mandible.....	115
- Craniosynostosis and abnormal head shape....	123
- Facial masses	
*Oral & cervicofacial masses.....	137
*Scalp cysts .....	153
- Fetal syndromes fetaturing CFA .....	158
<b>VI. Summary and conclusions.....</b>	<b>163</b>
<b>VII. References.....</b>	<b>167</b>
<b>VIII. Arabic summary.....</b>	<b>209</b>



# List of figures

*Figure 1 Anatomy of the face and cranium.*

*Figure 2 Anatomy of the nose.*

*Figure 3 Normal anatomy of the external ear.*

*Figure 4 External anatomy of the eye.*

*Figure 5 (a) Anterior view of the right orbit (b) A schematic illustration of the various parts of the human eye.*

*Figure 6 the major anatomic landmarks of the lips, mouth.*

*Figure 7 Components of the hard palate.*

*Figure 8 Components of the nasal septum.*

*Figure 9 The fetal skull.*

*Figure 10 Neural crest cells migration.*

*Figure 11 Derivatives of the pharyngeal arch cartilages.*

*Figure 12 Frontal views of the heads of human embryos from 4 to 8-10 weeks of age.*

*Figure 13 Maxillary processes and its components.*

*Figure 14 The formation of the palate.*

*Figure 15 Orofacial clefts.*

*Figure 16 early stage of ear development.*

*Figure 17 Auricle development*

*Figure 18 Schematic depiction of early eye development*

*Figure 19 sagittal sections of the eye showing successive developmental stages*

*Figure 20 Ossification sites of membranous skull bones.*

*Figure 21. Synotia.*

*Figure 22 Acquisition with specific 3D probes.*

*Figure 23 Adjusting the region of interest (ROI) box before acquisition.*

*Figure 24 Adjusting the rendering box after acquisition.*

*Figure 25 4D ultrasound.*

*Figure 26 VCI-A of the foetal face*

*Figure 27 Multiplanar display.*

*Figure 28 multislice display*

*Figure 29 (A) surface rendering; (B) transparent maximum mode. 43*

*Figure 30 Adjusting the threshold level.*

*Figure 31 Use of 'electronic scalpels.*

*Figure 32 Rendering artifact creating the false image of "hole in the head."*

*Figure 33 Motion artifact presented as “conjoined twins”*

*Figure 34 Movement artifact feigning a cleft lip right (white arrow).*

*Figure 35. Example for an examination protocol of the fetal face with two-dimensional ultrasound.*

*Figure 36 Schematic demonstration of the approach to the 3D ultrasound evaluation of the fetal face.*

*Figure 37 Coronal and oblique ultrasound views for the assessment of the fetal face.*

*Figure 38 Axial ultrasound views for the assessment of the fetal face. 55*

*Figure 39 Standard orientation of the fetal face.*

*Figure 40. Three-dimensional ultrasound: the maximum or transparent mode.*

*Figure 41 Embryonic and human facial development depicted in utero by 3DUS surface rendering during the first trimester.*

*Figure 42 Fetal profile at different gestational ages*

*Figure 43. Sagittal (A), axial (B, C), and coronal (D) views of the fetal head demonstrating the relevant details of facial anatomy.*

*Figure 44 Representative ultrasound images of a normal fetal face, demonstrating coronal section planes.*

*Figure 45 Representative ultrasound images of a normal fetal face, demonstrating sagittal section planes.*

*Figure 46 Ultrasound image (sagittal section) depicting the proposed landmarks to identify a true midsagittal profile view of the fetal face*

*Figure 47 Representative ultrasound images of a normal fetal face, demonstrating axial section planes.*

*Figure 48 Summary of the most important anomalies of the fetal facial profile.*

*Figure 49 .These images illustrate how the use of 3D rendering can aid in exact positioning with regard to the midline of the fetal face.*

*Figure 50 The possibility of simultaneous visualization of several features with 3D surface rendering.*

*Figure 51 It can be seen here how 3D sonography can help to visualize not only severe facial malformations in a realistic way, but also subtle dysmorphisms.*

*Figure 52 Precise evaluation of fetal behavioral expressions.*

*Figure 53 Schematic diagram showing anatomy of the palate and its various cleft constituents.*

*Figure 65 Anterior coronal 2D plane of the fetal face demonstrating unilateral cleft lip.*

*Figure 56 .Axial 2D planes of the maxilla in fetuses with facial cleft. 79*

*Figure 57. Three-dimensional surface analysis ultrasound images of cleft lip and alveolus.*

*Figure 58 .Three-dimensional ultrasound of cleft lip in surface mode and maximum mode*

*Figure 59 Bilateral cleft lip and palate with premaxillary protrusion 80*

*Figure 60 Midsagittal plane using bi-directional high definition (HD) Power Doppler flow.*

*Figure 61 the secondary palate by MRI*

*Figure 62 Reverse face view technique.*

*Figure 63 Reverse face view technique*

*Figure 64 Flipped face technique.*

*Figure 65 Axial underside view representation of the fetal palate 86*

*Figure 66 Tomographic ultrasound images of the secondary palate in the coronal plane.*

*Figure 67. A schematic illustration of the various parts of the human eye.*

*Figure 68. Hypertelorism and hypotelorism*

*Figure 69 Fetal MRI for the orbit.*

*Figure 70 The normal fetal eye*

*Figure 71 (A) Unilateral microphthalmia ,(B) Fetal cataracts*

*Figure 72 Persistent hyperplastic primary vitreous (PHPV)*

*Figure 73 A false-positive diagnosis of microphthalmia*

*Figure 74 Volume of the eye ball in a 25-week-old fetus.*

*Figure 75 Fetus with bilateral anophthalmia by 3D “reverse face” view technique.*

*Figure 76 Three-dimensional ultrasound images at 38 weeks' gestation in a case of unilateral anophthalmia.*

*Figure 77 Evaluation of the palpebral fissure shape.*

*Figure 78 A case of retina coloboma.*

*Figure 79. Multiplanar views of the dacryocystocele shows the characteristic location.*

*Figure 80 Three-dimensional scan in minimum surface mode*

*Figure 81. Tomographic sonographic imaging demonstrates the location and the relationship of the cystic mass with the surrounding anatomical structures.*

*Figure 82 (A-C) Axial and coronal T2-weighted prenatal single-shot, fast-spin echo magnetic resonance imaging of a fetus with bilateral dacrocystocele,*

*Figure 83 Visualization of the fetal ear by ultrasound.*

*Figure 84 Clear visualization of ear morphology by the surface rendering mode.*

*Figure 85 Low set ear demonstrated by ultrasound*

*Figure 86 Pre-auricular tags*

*Figure 87 Ultrasound images demonstrating appropriate examination of fetal nasal bone.*

*Figure 88 Fetal profiles at 12 weeks of gestation in a normal fetus, showing the nasal bone, and a trisomy 21 fetus, showing absence of the nasal bone.*

*Figure 89 Ultrasound images showing measurement of the frontomaxillary facial angle*

*Figure 90 Fetus with Down syndrome at 22 menstrual weeks. A midline sagittal view of the face clearly shows the absence of a nasal bone. The parasagittal view falsely displays what appears to be a nasal bone.*

*Figure 91 the effect of deviations from the exact mid-sagittal plane on the measurement of FMF angle.*

*Figure 92. Possibility of error when assessing nasal bones only on two-dimensional (2D) imaging demonstrated in a fetus with unilateral absence of a nasal bone.*

*Figure 93 Transverse views of the fetal nasal bones in the multiplanar mode of three-dimensional ultrasound.*

*Figure 94. 3D maximal rendering of three different conditions: (a) normal bilateral nasal bones; (b) absent nasal bone in a fetus with Down syndrome; (c) unilateral absence of a nasal bone*

*Figure 95. Subjective diagnosis of micrognathia*

*Figure 96 Examples of objective assessment of the mandible.*

*Figure 97. Three-dimensional multiplanar imaging is used to standardize orthogonal views of the face.*

*Figure 98 Pierre Robin sequence .*

*Figure 99 Pierre Robin sequence .*

*Figure 100 Three-dimensional rendered images of fetuses with syndromic micrognathia.*

*Figure 101. Three-dimensional rendering (maximum mode) of the normal fetal mandible at 20 weeks of gestation.*

*Figure 102. Caffey disease, 21.9 menstrual weeks.*

*Figure 103 Three-dimensional ultrasound of agnathia.*

*Figure 104 Major sutures involved in craniosynostosis.*

*Figure 105 Two-dimensional ultrasound of the cranial sutures and fontanelles.*

*Figure 106 2D images of craniosynostosis.*

*Figure 107 3D ultrasound reconstruction of the cranial sutures and fontanelles.*

*Figure 108 3DUS with transparent maximum mode demonstrating the closed coronal suture in a fetus with Apert syndrome.*

*Figure 109. Normal development of the metopic suture.*

*Figure 110 Three-dimensional maximum mode rendering of the anterior fontanelle.*

*Figure 111 Apert syndrome.*

*Figure 112 Pfeiffer syndrome. 2D*

*Figure 113 Pfeiffer syndrome, 3D surface mode*

*Figure 114 Pfeiffer syndrome type 2 with cloverleaf skull. Examination of the hands reveals bilateral abducted broad thumbs with possible partial duplication of the first phalanx.*

*Figure 115 Use of 3DUS in differentiating craniosynostosis from severe fetal molding.*

*Figure 116 Value of MR imaging in prenatal diagnosis of craniosynostosis.*

*Figure 117 Ex Utero Intrapartum Treatment (EXIT) Procedure.*

*Figure 118 Sagittal single-shot fast-spin echo (SSFSE) T2-W image depicts a upper airway in a fetus.*

*Figure 119 Fetal magnetic resonance image demonstrating an obstructing cystic intraoral mass.*

*Figure 120 A case of congenital epilus at 32 weeks' gestation.*

*Figure 121 Interactive, multidirectional 3DUS displays and virtual assessment of the relationship between the fetal oral tumor and route of intubation.*

*Figure 122 3D sonographic view depicts a large stalk of the main mass*

*Figure 123 3D surface renderings of the fetal face showing the extra-oral component of a mass protruding from the fetal mouth.*

*Figure 124 Prenatal surface-rendered coronal image (left) of the fetal face with threshold modification.*

*Figure 129 Ultrasound examination using 3-D multislice view of the fetal face at 35 weeks' gestation with an exophytic mass.*

*Figure 126 A case of a small palatal teratoma diagnosed at 37 weeks by 3D sonography.*

*Figure 127 Tongue teratoma at 30 weeks' gestation.*

*Figure 128 Evaluation of an oral mass (a congenital epulis)*

*Figure 129 A giant nuchal hemangioma. 4D and 3D ultrasound reveal a lobulated subcutaneous mass.*

*Figure 130 2D and 3D images of a large lymphangioma that impairs the airway passages.*

*Figure 131. (a) Conventional two-dimensional ultrasound of a scalp cystic mass (b) Three-dimensional multiplanar mode with surface-rendered imaging.*

*Figure 132 Three-dimensional multiplanar views of the fetal brain pinpoint the location of the small bone defect*

*Figure 133. Transvaginal sonogram of the scalp cyst.*

*Figure 134 Three-dimensional power Doppler angiograms of the meningocele.*

*Figure 135: Frontonasal dysplasia.*

*Figure 136 Noonan syndrome.*

*Figure 137 De Lange syndrome.*

*Figure 138 Binder syndrome.*

*Figure 139 The "Greek warrior helmet" appearance.*

*Figure 140 Fetal facial profile in Seckel syndrome.*

*Figure 141 Harlequin ichthyosis.*

*Figure 142 Achondroplasia.*

*Figure 143 Hemifacial microsomia.*

# List of tables

<i>Table 1 Derivatives of Facial Components.</i>	<i>14</i>
<i>Table 2 Steps required in 3D ultrasound.</i>	<i>29</i>
<i>Table 3 Assessment of the fetal face by 3D US in the first trimester.</i>	<i>54</i>
<i>Table 4 Assessment of the fetal face by 3D US in the second and third trimesters.</i>	<i>55</i>
<i>Table 5 Normal appearance of fetal facial features on ultrasound in the coronal section planes</i>	<i>58</i>
<i>Table 6 Normal appearance of fetal facial features on ultrasound in the mid-sagittal section plane</i>	<i>59</i>
<i>Table 7 Normal appearance of fetal facial features on ultrasound in the axial section planes</i>	<i>60</i>
<i>Table 8 Normal appearance of fetal facial features on ultrasound in the parasagittal section planes</i>	<i>61</i>
<i>Table 9. Brief overview of syndromic craniosynostoses.</i>	<i>124</i>
<i>Table 10 Information provided by fetal US vs fetal MR imaging prior to an EXIT procedure.</i>	<i>141</i>

# List of abbreviations

2D .....	Two-dimensional
2DUS.....	Two-dimensional ultrasound
3D.....	Three-dimensional
3DUS.....	Three-dimensional ultrasound
4D .....	Four-dimensional
ACS.....	Acrocephalosyndactyly
BOD.....	Binocular distance
CFA.....	Craniofacial anomalies
CI .....	Cephalic index
CLP.....	Cleft lip and palate
CL±P .....	Cleft lip with or without cleft palate
CL+A.....	Cleft lip and alveolus
CP.....	Cleft palate
DS.....	Down syndrome
EXIT.....	Ex utero intrapartum treatment
FGFR.....	Fibroblast growth factor receptors
FMF (group) .....	Fetal Medicine Foundation
FMF (angle) .....	Frontomaxillary facial angle
HA .....	Hyaloid artery
IOD .....	Interocular distance
IFA .....	Inferior facial angle
MSV .....	Multislice view
NB .....	Nasal bone
NT.....	Nuchal translucency
OD .....	Ocular diameter
PHPV.....	Persistent hyperplastic primary vitreous
PRS .....	Pierre Robin sequence
PT.....	Prenasal thickness
ROI .....	Region of interest
TUI .....	Tomographic ultrasound imaging
VCI .....	Volume contrast imaging
VOCAL.....	Virtual organ computer aided analysis

