CRANIOFACIAL ANOMALIES AMONG NEW BIRTHS AT TWO HOSPITALS IN NAIROBI

DR. ODHIAMBO ATANASIAS, BDS (NBI).

A DISSERTATION SUBMITTED IN PART-FULFILLMENT OF THE REQUIREMENTS FOR THE DEGREE OF MASTER OF DENTAL SURGERY IN ORAL AND MAXILLOFACIAL SURGERY

UNIVERSITY OF NAIROBI

AUGUST 2007

DECLARATION

I, Dr. Atanasias Odhiambo certify that this dissertation is my own original work and has not been presented for a degree in any other university.

Signed _____

Dr. A. Odhiambo, BDS (U.O.N)

Date 20/9/07

APPROVAL

This dissertation has been submitted to the University of Nairobi for examination with our approval as supervisors:

PROF. M.L. CHINDIA, BDS, MSc, FFDRCSI.
Chairman and Associate Professor,
Dept. of Oral and Maxillofacial Surgery, Oral Pathology and Oral Medicine,
School of Dental Sciences, University of Nairobi.
Signature: ..... Date ..... 20.9.2007.

DR. MUIA NDAVI, MBCHB, MMED (OBS/GYN), MSc.Epid, FHBR, DLSHTM.
Senior Lecturer,
Department of Obstetrics and Gynaecology,
School of Medicine,
University of Nairobi.
Signature: .....Date: 21/9/07.....

DR. F.G. MACIGO, BDS, MPH, PGD-STI.
Senior lecturer,
Department of Periodontology / Community and Preventive Dentistry,
School of Dental Sciences, University of Nairobi.
Signature: ..... Date: 20/9/2007

DR. F. WERE, MBCHB, MMED (PAEDS), MD.
Senior lecturer,
Department of Paediatrics and Child health,
School of Medicine, University of Nairobi.
Signature: .....Date: 20/9/07

DEDICATION

This work is dedicated to the innocent human beings whose lives are shattered or claimed by malformations.

ACKNOWLEDGEMENT

I wish to thank the leadership of the Kenya Navy Headquarters for the financial support. Similarly, I thank my supervisors Prof. M.L. Chindia, Dr. Muia Ndavi, Dr. F.G. Macigo and Dr. F. Were for their encouragement and guidance throughout the study. The statistician, Ms. Alice Lakati of Kenya Medical Training College, Nairobi helped me a great deal with data analysis.

I acknowledge the co-operation of the administrations and all the staff of Kenyatta National Hospital and Pumwani Maternity Hospital who were working in the study areas during the study period especially in the labour wards, new born units, postnatal wards and the mortuaries. Lastly, I salute my wife and children for being there for me through out and above all, the almighty God for invigorating me time and again.

TABLE OF THE CONTENTS

LIST OF APPENDICES

LIST OF FIGURES AND TABLES

Page

ABBREVIATIONS

CFAs	*Craniofacial anomalies*
KNH	Kenyatta National Hospital
PMH	Pumwani Maternity Hospital
FSB	Fresh stillbirth
MSB	Macerated stillbirth
NBU	New-born unit
SVD	Spontaneous vertex delivery
CS	Caesarean Section
PS	Preauricular sinus
PT	Preauricular tag
NR	Not recorded, invalid entry, year of birth not known.
WBAs	Whole-body anomalies (any part of the body).

DEFINITION OF TERMS

1. **Birth defect** refers to an abnormal development originating prenatally whether present at birth or expressed later.

2. **Congenital malformation** refers to gross structural anomalies, internal or external, either severe or mild, resulting from a localized error of morphogenesis, which may be caused by environmental or genetic factors.

3. **Major (severe) congenital** anomaly may be fatal or handicap the patient throughout life while **minor anomalies** such as skin tags do not handicap the infant.
4. **Anomalies and malformations are terms which will be used interchangeably in this dissertation.**
5. **Genetic illness** implies the passing of abnormal genes or chromosomes from one or both parents to the child.

6. **Acquired** (environmental factors) diseases arise sporadically through genetic mutations or direct assault on the foetus by hazardous factors with parental genes having little or no role in the child's illness.

7. The term **Syndrome** refers to a recognised pattern of malformation, such as the Down's syndrome, presumably having a common aetiology.

8. **Clinically recognizable** in this study refers to visible and or palpable anomalies such as clefts and tags. Percussion and auscultation were not done. Lesions such as congenital tumours which need histological examination and anomalies not involving the oral and craniofacial region were grouped together as 'others'.

ABSTRACT

BACKGROUND: The pattern of congenital oral and craniofacial anomalies (CFAs) in the Kenyan population remains unknown. The few studies on whole-body congenital anomalies in Kenya and other African countries have suggested that the pattern of anomalies may be significantly different from those reported in other races worldwide. Kenyan studies have reported on anencephaly, hydrocephalus, encephalocoeles and cleft lip and palate with no mention of the other oral and craniofacial anomalies such as astomia, aglossia,microtia, or preauricular sinuses and tags. Therefore, the pattern of occurrence of these anomalies needed to be established.

OBJECTIVE: To describe the pattern of occurrence of CFAs at two hospitals in the city of Nairobi.

DESIGN: A descriptive cross-sectional study.

SETTING: Kenyatta National Hospital (KNH) and Pumwani Maternity Hospital (PMH).

SUBJECTS AND METHOD: The study was done from November 2006 to March 2007. All mothers who delivered at the two hospitals were consented for interview and examination of their babies. All births were physically examined within 48 hours by the investigators for any anomalies from head to toe and intra-orally using a clinical examination form. Anomalous infants were classified for type, location and magnitude of anomalies. Data were analysed to determine the association of these anomalies with ages of mothers, gender, weight, birth order, mode of delivery and birth status of the babies using the statistical package for social science (SPSS) software version 12.0 and EpiInfo package. Descriptive and inferential statistics were done using the X^2 and Fisher exact tests.

RESULTS: During the study period 7989 babies were born in the two hospitals among whom 4264 (53.5%) were males and 3721(46.6%) were females and 4(0.1%) had ambiguous external genitalia. Total whole body anomalies were 256 (3.2%) among all births. Anomalous males were 142 (1.8%) and females

were 110 (1.4%) of all the neonates. The most common single anomaly was preauricular sinus constituting 34(4.3/1000) of the total births, followed by extra digits at 22(2.8/1000), then talipes at 20(2.5/1000) of total births. Total CFAs were 146, comprising 57.3% of whole body anomalies and 1.8% of the total livebirths. CFAs were more common in female livebirths (1.4%) than the male (1.0%) livebirths. However, a total of 23.3% of stillbirths had CFAs, with lesions manifesting more commonly in the males (16.5%) than the females (6.8%). The commonest CFA was preauricular sinus 0.4% (4.3/1000births) followed by hydrocephalus at 0.19% (1.9/1000) then cleft lip and palate and preauricular tags at a prevalence of 1.6/1000 and 1.5/1000 respectively. Anomalies were significantly common in the first birth order and tapered off steeply past the fifth born. Out of a total of 5930 spontaneous vertex deliveries (SVDs) 148 (2.5%) had anomalies and out of 2059 caesarean sections (CSs) 103 (5.0%) had anomalies. Major CFAs occurred at 0.6% (6.1/1000) in the livebirths and at 65.3% of the stillbirths while single minor anomalies occurred at a rate of 1.1% (11.3/1000) of the total births. Multiple major anomalies were common in stillbirths (85.7%).

CONCLUSION: The commonest CFAs were preauricular sinus, hydrocephalus and clefts of the lip and palate. The anomalies were significantly common in the first and second birth-order, low birth weight babies, in babies delivered via caesarean section and in male stillbirths, comparing well with the findings by other investigators in the literature. Minor CFAs were significantly associated with other clinically recognizable anomalies all over the body. Major CFAs were fatal in more than half the time within 48 hours perinatally.

CHAPTER 1

1.1. INTRODUCTION

Generally congenital craniofacial anomalies (CFAs) are rare.[1] Among them, cleft lip and or palate, occurring at the rate of 0.06-2.13/1000 live-births are the most common.[2,3] The highest incidence of cleft lip and palate has been reported in the Indian tribe of Montana(1:276), followed by Oriental groups (1:500) and the least affected are the Negroid population (1:2000).[4] Most congenital anomalies of the head originate during the transformation of the pharyngeal apparatus into adult structures.[5] Single minor anomalies occur in 14% of newborns and should be recognised as the possible indicator of an associated serious malformation.[5,6,7] Major congenital malformations are found in 2% of live-births, 22% of stillbirths; and there is an increase in the rate of congenital malformations in consanguineous marriages.[6] Malformations due to mutant genes vary from about 0.5-0.8/1000 births, while about 6% of all serious malformations in live-born infants are associated with major chromosomal anomalies, a figure which rises when the stillbirths are karyotyped.[7,8]

Environmental causes of congenital anomalies include irradiation, drugs (e.g. anticonvulsants), nutritional extremes (folic acid and vitamin A), hyperthermia or hypothermia. Vitamins and folic acid supplements taken prior to conception reduce the incidence of neural tube defects, while certain drugs such as methotrexate and steroids cause these defects in 1 to 25% of pregnancies if given in the first 4 weeks of development.[7,8,9] About 2% of all malformations

are caused by infectious diseases, 1.4% due to diabetes mellitus and less than 1% due to other diseases implying that 3.5% of all congenital malformations are due to maternal illness while idiopathic causes comprise about 85% of all anomalies, 1% of the patients manifest additional birth defects (syndromes).[8,10] Maternal age most involved in congenital anomalies is between 20 and 35 years and the pregnancies affected are mostly breech presentations and often the first born. A higher frequency of major anomalies occurs in multiple births than single births and males have an excess of malformations over females.[11,12]

1.2. LITERATURE REVIEW

Classification of OCFAs is usually based on new theories but it always remains controversial.[1] Anatomical classification includes:

I. ***Oral anomalies***: These include facial clefts, macrostomia, microstomia, astomia, ageniocephaly, micrognathia, macrognathia, agnathia, short upper lip, long upper lip, white spongy nevus, leukoedema, hypertrophy of submandibular and sublingual salivary glands, mucocoeles, branchial cyst, dermoid cyst/epidermoid cyst, cystic hygroma, congenital ranula, otocephalia, coloboma, pits, fistulae, whistling face deformity, crying face deformity, neonatal epulides, idiopathic gingival fusion, alveolar cysts/gingival cysts, alveolar lymphangioma, median alveolar notch, natal teeth, tongue anomalies including aglosia, bifid/lobulated tongue, ankyloglosia, accessory tongue, scrotal/fissured tongue, cyst in the tongue.

II. ***Nasal anomalies***: Nasal aplasia, aplasia with proboscis, nasoschizis, nasal duplication, hypoplastic nose, broad/flat nasal bridge, choanal atresia, small upturned nose.

III. ***Ear (aural) anomalies***: Preauricular sinus, fistulae/cyst, prominent ear, adherent ear, constricted ear, anotia, microtia, aural appendages (tags).

IV. ***Eye anomalies***: Cyclopia, ethmocephaly, synophthalmia, microblepharon, microphthalmia, anophthalmos, Cryptophthalmia, Congenital aphakia, blepharoschizis, blepharoptosis, microblepharon, euryblepharon, ankyloblepharon, epiblepharon, epicanthus, canthal dystopia, canthoschizis, blue sclera and persistent iridopupillary membrane.

V. ***Cranial anomalies***: acrania, anencephaly, cranium bifidum, microcephaly, macrocephaly, congenital hydrocephalus.

VI. ***Others:*** congenital alopecia, Peutz-Jeghers Syndrome, hypopigmentation, neurofibromatosis, absence of skin, facial nerve palsy, facial hemi-hypertrophy and the other body systems.

ORAL ANOMALIES

The mouth and perioral region are best observed with the infant both at rest and crying. Lateral nasomaxillary clefting occurs between the lateral nasal and maxillary processes *(naso-ocular clefts)*, the medial nasomaxillary clefting occurs between the medial nasal and maxillary process *(cleft lip)*,the intermaxillary cleft would be between the palatine processes *(cleft palate)*, while the maxillomandibular clefting occurs between the maxillary and mandibular processes *(macrostomia)*. Macrostomia is commonly associated with auricular tags and fistulae and it may be bilateral or unilateral extending from the angle of the mouth up to the masseter muscle or in severe cases up to the tragus of the ear. Its incidence varies from 1 in 100 -330 births.

It commonly occurs on the left side and may be associated with harelip and cleft palate.[1,7,13]

The Philippines have a birth prevalence of 1.9/1000 live-births for cleft lip and palate, a recurrence rate in siblings for nonsyndromic cases of 23 per 1000 for cleft lip with or without cleft palate and 14 per 1000 for cleft palate only. The percentage of clefts associated with multiple anomalies is 21% at birth, with a higher prevalence in boys than girls.[14,15] Iregbulem (1982)[16] in a Nigerian study found the incidence of clefts of the lip or palate or both to have been 1 in 2,703 live births and the right and left sided lip clefts occurred in equal proportions although in complete clefts, the left sided lesions were predominant with a slight male predorminance.[17] Lack of cell degeneration seems to be one of the main causes of clefts[7] but they may also occur due to mechanical obstruction of the palatal shelves by the tongue, insufficient growth of the palatal processes or failure of capillary formation in the marginal areas of the distribution of the arteries of the face leading to necrotic zones associated with amniotic adhesions. Cleft lip and palate occur at 1-3/1000 live births in African neonates and 1 in 600-800 in whites.[18]

The risks of clefts in stillbirths and abortions is 3 times greater than in live-births.[17] The incidence of posterior cleft palate is similar in different races whereas the incidence of cleft lip with or without cleft palate varies with races (most common among the Asians and less common in the Negroids). Cleft palate may involve the soft palate only, isolated cleft hard palate (1:2200), both hard and soft palate, the submucosa of the palate (1:1200), bifid uvula (1:100). Lower lip clefts are mostly median while the upper lip clefts may be midline (true harelip), unilateral (commonly on the left side) or bilateral and may be

associated with clefts of the alveolar process or palate.[19] Clefting is highest on the left, among those in low socioeconomic class and in males than females (male: female=63%:37%). Isolated clefts are more in females than males 66%:34%, harelip with or without cleft palate (1:2500 births) occur mostly in males at 70% to 30%. Simple harelip is rarer than the harelip-cleft palate complex (cheilognathopalatoschisis) and unilateral clefts of the lip are more common than bilateral ones.[20]

Microstomia is a small mouth, while *astomia* is union of the upper and lower lips. Microstomia occurs in maternal hypervitaminosis A and in syndromes such as trisomy 18, 13-15, Hallerman-Streiff, Freeman and Sheldon.[1,21] *Ageniocephaly* (chinlessness) presents with an underdeveloped mandible, tongue and microstomia.[19] *Micrognathia/Macrognathia* refers to decreased and increased jaw size respectively. Micrognathia occurs in vitamin B_2 deficiency.[19] *Agnathia* (partial or total) refers to the absence or agenesis of the maxilla or mandible.[1] Unilateral maxillary agnathia is due to failed development of one maxillary process. A *Short upper lip* occurs as a symptom in the popliteal pterygium and the Ellis–Van Creveld Syndromes. An *elongated upper lip* is described in the de-Lange syndrome and in craniocarpotarsal dystrophy.[21] In the oral mucosa *White spongy nevus* may be present at birth as a bilateral wide area of the oral mucosa covered by white thick patches which appear folded or corrugated on the mucosa of the cheek, lips, tongue, gingiva or floor of the mouth. *Leukoedema* is a variant of the normal oral mucosa with diffuse filmy striaform, opalescence of the buccal mucosa. If the mucosa is stretched, the leukoedema disappears. It occurs in blacks at 43% and 11% in the whites.[13]

Hypertrophy of the submandibular and sublingual salivary glands may be associated with aglossia. A report of enlarged sublingual glands in a child with trisomy 18 exists.[10] Congenital *mucocoeles* are apparent after birth and may interfere with mouth closure. Most are retention cysts due to trauma of the duct or gland causing atresia or extravasation of saliva leading to an extravasation mucocoele. Congenital atresia of the submandibular and sublingual ducts has also been considered as a possible cause.[22] The neck and floor of the mouth anomalies include the *branchial cyst, dermoid cyst/epidermoid cyst, cystic hygroma and congenital ranula*.[23]

Mandibular anomalies occur when the arch and the body are reduced in all dimensions with the backwardly positioned chin and in its severest form it is called *otocephalia*.[1,19] Intermandibular anomaly varies from a small *coloboma* of the lower lip to a complete cleft lip, mandible and tongue extending back and downwards between the genioglossus muscle and sternal notch.[7] The midline mandibular cleft is believed to occur due to pressure from the adjacent enlarged heart which begins to beat before the fusion of the mandibular midline.[24] Congenital *pits* and *fistulae* occur in the lower lips at 1in 2billion live-births but lateral lip pits are common in negroes.[25] Upper lip fistulae occur at the junction of the globular and maxillary processes or in the middle at the origin of the labial fraenum.[26] About 70–80% of the patients with lower lip pits may have cleft lip and palate or isolated cleft palate.[19]

Alveolar and gingival anomalies include *neonatal epulides* and congenital *fusion of the gums*.[13] Congenital epulides are common in the maxillary incisor region and are commonest (female: male is 8-10:1) in females.[13,27] *Alveolar /gingival cysts* of the newborn have an

incidence of 26-53% in whites and 11-40% in black neonates. They are due to cystic degeneration of the remnants of the dental lamina and are found on the alveolar crest lingually or buccally.[28] *Alveolar lymphangioma* presents as a blue-domed, fluid-filled lesion in the posterior aspect of the alveolar crest; buccal or lingual and is not associated with an unerupted tooth; and since the alveolar crest is devoid of salivary gland tissue, this is not a mucous retention cyst. The lymphangiomas are found in 3.7% of live-births in blacks. Palatal cysts or raised fluid filled lesions of the palatal mucosa in neonates are common with 58 to 64% of newborns having yellow-white elevated cysts located or adjacent to the midpalatal raphae or at the junction of the hard palate. It can be one or 3 to 6, less than 1mm in diameter and are common in white than black neonates. They are called Epstein pearls if located within the fusion of the posterior palatal segments and Bohn's nodules if adjacent to the mid palatal raphae or along the junction of the hard and soft palate.[28]

The *median alveolar notch* (common in the maxilla) may be associated with cleft lip without cleft palate. The cause of the median alveolar notch is attributed to the labial fraenum tension on the maxillary ridge. It has a frequency of 20-26% and is manifest more in black neonates.[28] *Natal teeth* are often paired, present at birth, and are common in the mandibular central incisor region[6] though other investigators report their predilection for the molar area.[28] The incidence of natal teeth is 1 in 700-6000 births, 8% to 46% have a positive family history[27] and are commoner in females than males occurring in 25-30% of the Ellis Van Creveld Syndrome.[29]

Tongue anomalies include *bifid(glossoschisis), lobulation, reduced size, absence(aglossia), ankylosis,[28] an accessory tongue, lingual cysts,*

fistulae, fissured and scrotal tongue.[13] *Glossoschisis* is due to incomplete fusion of the distal tongue buds.[8] *Lobulated tongues* are of normal size and shape, but attached to them are much smaller lobe(s).[26,30] *Ankyloglossia* occurs in 1.7% of neonates with no sex or racial predilection and may be inferior (lower jaw) or superior (attached to the palate), may be isolated or occur in a syndrome.[19,28] Inferior ankyloglossia occurs in 1 in 300 American infants.[7,13] *Congenital macroglossia* is due to overdevelopment of the muscular part of the tongue or Beckwith-Wiedman syndrome and may also be seen in generalized muscular hypertrophy of the rest of the body. Hemi-hypertrophy of the body or the face (only ½ tongue is involved) is seen also in cretinism, infantile myxoedema, lymphangioma and small tumour masses.[13]

OCULAR ANOMALIES

Cyclopia refers to a deformed single median eye, enclosed in a single orbit in the middle of the forehead due to translocation of chromosomes[18] leading to improper development of the frontonasal process. In *Ethmocephaly,* facial malformations resemble those in cyclopia but the orbits are separated from each other. *Synophthalmia* is fusion of the eyes (partial or complete).[1,7] The eyelids may be reduced in size (*microblepharon*), while ptosis and hirsutism of the upper eyelid may be observed.[7] *Microphthalmia* is used in cases in which the presence of some ocular development defect may be established by clinical means. *Anophthalmos* (congenital absence of all eye tissues) and microphthalmos may be found in combination with many other anomalies.[2,7] Microphthalmia may be inherited or due to *Rubella, Herpes simplex viruses* or *Toxoplasma gondii* infections intrauterine.[8,9] *Cryptophthalmia* refers to complete fusion of the upper and lower eyelids, absent eyelashes, eyebrows may be partially

defective or absent; and the position of the eyeball is indicated by a swelling and eye movement can sometimes be seen.[1,7] It is an autosomal recessive condition. *Blepharoschizis* (coloboma) can be a simple notch in the eyelid rim or a complete absence of the eyelid (ablepharon) and may be associated with Treacher Collins-Franceschettiklein syndrome which expresses an underlying malar deficiency and[1] is common in upper eyelids.[7]

The incidence of congenital coloboma ranges from 0.5 in Spain, 1.4 in France, 2.6 in the USA to 7.5 per 100,000 births in China. Coloboma is found in 3.2-11.2 of blind children worldwide. *Blepharoptosis* (congenital ptosis) is due to abnormal development of the levator palpebrae superioris muscles or abnormal superior division of the occulomotor nerve.[7] *Microblepharon refers to a shortened eyelid in all* directions usually associated with microphthalmia, micro-orbitism and *blepharophimosis. Euryblepharon is an enlarged palpebral aperture* due to deficiency of the palpebral skin in vertical dimension and *ankyloblepharon* is fusion of the eyelids which may be partial or total while *epiblepharon* refers to the absence of palpebral groove, due to mal-development of palpebrotarsal insertion of the muscle levator.[1,7] *Epicanthus* refers to bilateral and symmetrical skin folds running in a vertical direction at the naso-orbital angle overlapping in the severest form the medial palpebral ligament.[1] *Canthal dystopia* implies an abnormal position of the medial or lateral canthus, like the mongoloid slanting and other syndromes while *canthoschizis* is clefting of the lateral canthus which may be associated with scleral dermoid or lipoma or be part of the Goldenhar's syndrome.[1] *Persistent iridopupillary* membrane occurs due to incomplete resorption of the iridopupillary membrane leaving small strands of connective tissue over the pupil.[2] The *blue sclera* may be present in normal or abnormal

individuals such as *osteogenesis imperfecta, osteopetrosis, Marfan syndrome, foetal ricketts* and *Ehler Danlo's Syndrome*.[12]

NASAL ANOMALIES

Nasal aplasia indicates unilateral malformations while the word arhinia is used when both halves are absent. The incidence of aplasia with proboscis is less than 1:100,000 newborns.[1] *Congenital choanal atresia* may involve the anterior or posterior choana.[31] *Nasoschizis* (clefts) ranges from a minor notch of one or both alae to absence of nostrils and nasal bone often associated with other facial defects.[1] *Nasal duplication* ranges from a supernumerary nostril in an otherwise normal nose to duplication of the upper face (diprosopia). The supernumerary nose is usually the medial one and may end blindly, be stenotic or open into a nasal cavity. It may be bilateral, unilateral, single or in combination with other facial anomalies.[1,7]

EAR ANOMALIES

The incidence of external ear anomalies is about 1% with microtia being 1.69% and preauricular sinus ranging from 50-80%.[1,7] *Microtia* which arises due to suppressed development of auricular hillocks refers to a severely displastic or disorganised external ear and may serve as an indicator of associated anomalies such as atresia of the external acoustic meatus and the middle ear anomalies. Type I microtia has a small auricle that is deformed but has the essential structures, type II appears as a curving elevation representing a deformed helix while type III presents only primordial hillocks.[8,25] Non-syndromic cases occur at 1.69 per 10,000births.[18,25] Unilateral microtia occurs commonly on the right (86.5%) and more in males, least in whites, increased in mixed races; and there is an increased risk at parity 4+(standardised for maternal age). The canal is often missing and in many cases a

hereditary pattern is present as autosomal dominant or recessive. Little is known of exogenous causes of microtia but implicated factors are isotretinoin and thalidomide.[19,32] Severe microtia may be associated with facial paralysis due to the involvement of the facial nerve in its canal and involves the mandible in 50% of patients.[7,8] Minor anomalies of the auricles may serve as indicators of a specific pattern of congenital anomalies such as abnormality in shape and low-set in infants with chromosomal syndromes such as trisomy 18.[8]

Aural appendages (tags) occur due to the development of accessory auricular hillocks, are anterior to the auricle, mostly unilateral and are the most frequently encountered malformations. For nonsyndromic cases the general rate is 17 per 10,000births; 13.66/10,000 in whites and 19.10/10,000 in blacks. The appendages may have narrow pedicles which may contain cartilage.[7,33] *Fistulae, sinuses* and *cysts* are defects of pharyngeal arches due to incomplete obliteration of the related embryonic spaces with the occurrence of internal or external openings. Fistulae and sinuses are derived only from the second cleft and pouch defects.[2] The incidence of *preauricular fistulae among* African newborns is 5.2% of females, 3.6% of males: among the European population it is 0.9% and 10% among the Asians.[18,25] *Duplication of the external auditory* canal has also been reported, and *atresia* of the external auditory canal; and *auricular hypoplasia* do occur. Atresia occurs due to failure of the meatal plug to canalize and has no race or sex predilection.[25] Embryological basis of sinuses is related to abnormal development of the auricular hillocks, defective closure of the dorsal part of the first pharyngeal groove and some represent ectodermal folds sequestered during the formation of the auricle. Sinuses are familial and frequently bilateral.[7]

CUTANEOUS ANOMALIES

Congenital alopecia occurs due to failure of hair follicles to develop, or due to follicles producing poor quality hair. Random patches of white hair may occur in families or be sporadic. [18,25] *Absence of skin* (common in the scalp) due to small areas where skin fails to form gives the appearance of ulcers.[7]

CRANIAL ANOMALIES

Acrania refers to the absence of the calvarium. It is associated with an extensive defect of the vertebral column, meroanencephaly or anencephaly (partial absence of the brain). *Cranium bifidum* refers to a bony defect in the cranium which occurs about once in every 2000-5000 births[8] for occipital encephalocoeles while frontal encephalocoele is 1:35,000 with females being more affected than males (2:1).[34] *Microcephaly* (a normal sized face but the calvarium is very small)[10] is due to deficient neural tube development into the brain due to diverse factors including hereditary, infective, irradiation or chemical (maternal alcoholism) factors. This could be obvious or may need head circumference measurement.[5] *Macrocephaly* is an abnormally large head.[8] *Congenital hydrocephalus* (incidence is 0.4-0.8/1000 live and stillbirths) is caused by a blockage in the flow of cerebrospinal fluid (CSF), overproduction of CSF or under-absorption of CSF. Congenital aqueductal stenosis is transmitted by an x-linked recessive trait but is mostly due to foetal infection (*Cytomegallovirus, Toxoplasma gondii*) or prematurity associated with intraventricular haemorrhage leading to the obliteration of cisterns or arachnoid villi.[8,25]

1. 3. RESEARCH PROBLEM

Major congenital anomalies are amongst the leading cause of neonatal mortality, they contribute substantially to chronic disease morbidity, profoundly affect families and their management is expensive and long-term. Minor anomalies may be unwanted, cosmetically disfiguring and may be a sign of internal anomalies; hence the need to know their pattern of occurrence in the population. However, a review of the literature revealed a paucity of Kenyan data on specific congenital oral and craniofacial anomalies[20,37] necessitating this study.

1.4. JUSTIFICATION OF THE STUDY

This research adds to the existing body of knowledge, serves as a pioneer data-base on anomalies of the head and neck in Nairobi and the information obtained enables the counselling of parents with anomalous children using Kenyan-specific data.

1.5. OBJECTIVES

1.5.1 BROAD OBJECTIVE

To determine the occurrence of clinically manifest congenital oral and craniofacial anomalies among new births in Kenyatta National Hospital (KNH) and Pumwani Maternity Hospital (PMH), Nairobi.

1.5.2 SPECIFIC OBJECTIVES:

1. To describe the frequency of occurrence of oral and craniofacial birth anomalies.
2. To determine the pattern of site occurrence of the clinically identifiable birth anomalies.
3. To determine some of the socio-demographic factors associated with CFAs.
4. To correlate the Nairobi occurrence of CFAs with others in the literature.

CHAPTER 2

MATERIAL AND METHODS

2.1. Study area: This survey was done in the two largest government delivery centres, KNH and PMH in Nairobi.

2.2. Study population: All mothers who delivered and their babies.

2.3. Study Period: November 2006 to March 2007.

2.4. Study Design: This was a descriptive cross-sectional study of incidence and distribution of clinically manifest CFAs at birth.

2.5. Study Variables:

I. ***Dependent Variables***

.Presence of anomaly

II. ***Independent Variables (Demographic Characteristics)***

. Age of mother . Live or Still-birth . Mode of presentation

.Gender of baby .Weight of baby . Baby's birth order

. Mode of delivery

2.6. Sample Size: This was a descriptive cross-sectional study to determine the range and pattern of occurrence of clinically manifest CFAs among babies at birth. The sample size was calculated using the Fisher et al. (1998) formula for population studies. Single minor anomalies occur among 14% of newborns. Major anomalies are found in 3% of live-births and 22% of stillbirths. The sample size was calculated using the prevalence of single minor anomalies (14%) since these anomalies are the ones which have been widely reported and also due to time and resources available to the researcher.

$$n = \frac{Z^2_B P(1-P)}{d^2}$$

Where;

n= required sample size (number of anomalies).

p= prevalence of single minor anomalies at birth (live and stillbirths), (p= 14%).

d= precision of the study (at 5%).

Substituting the above values in the formula at the 95% confidence interval:

$n=\frac{1.96^2 \times 0.14 \times 0.8}{0.05^2}=185$ anomalies.

Therefore, the desired sample size was 185 anomalies.

2.7. SAMPLING PROCEDURE

All mothers who delivered and the babies delivered during the study period were included.

2.7.1. Inclusion Criteria

1. All births at 20-weeks or more gestation and/ or at least 500g birth-weight during the study period.
2. Mothers who consented to participate in the study.

2.7.2. Exclusion criteria

1. Births below 500g in weight or less than 20-weeks gestation because of their poorly defined anatomic features.
2. Mothers who declined to consent.
3. Non-Kenyan mothers.

2.8. DATA COLLECTION PROCEDURE AND TOOLS

All women admitted for delivery were requested to consent for interview and examination of their babies within 48 hours of admission. Interview and examination form (Appendix I) was used to document the demographic data and record findings from systematic examination of all births done by the midwives who had been trained by the principal investigator (PI) on how to complete the forms and head to toe examination of the infants to elicit any anomalies. The registrars, medical officers, interns and the nurses in the labour, maternity and the newborn units, and the mortuary attendants were informed of the study and requested to assist the investigators.

The anomalies were classified into specific structures involved, major or minor and whether single or multiple. Each centre was manned day and night. The PI visited the study sites daily and during each visit he randomly picked the completed interview schedules at each centre and re-interviewed the mothers to find out whether the assistants were standard in their interview style and courteous to the mothers. He also examined all births which were present at that time. A pilot study was done by the investigator in the presence of the assistants and alterations were made to the interview schedules to ambiguous questions.

Whenever a case was delivered, the investigator was summoned via his mobile phone by the assistants or matron in-charge of the study site. Any infant with anomaly transferred to the newborn unit before examination was examined in the newborn unit, while any stillbirth transferred to the mortuary before examination was followed to the mortuary by the investigator for examination plus photography where indicated. In this research, each malformation was be counted once,

such that if an infant had both cleft lip and encephalocoele, it thus entered both classes for the tabulation of the number of infants with each anomaly that were born in the population. When an infant had a group of malformations that constituted a known syndrome or chromosomal disorder, then the syndrome was taken as the diagnosis but all the separate anomalies were also entered in the different classes. Parents with questions on malformations were counselled and educated by the investigators. Referrals for further management were done according to the laid down referral rules of each hospital.

2.9. DATA MANAGEMENT

Data were entered, coded and cleaned using frequencies and missing values counter-checked and corrected. Descriptive statistics were carried out for all continuous and categorical variables. X^2 and Fisher exact tests were used to determine association between occurrence of anomalies with ages of mothers, gender, weight, birth order, mode of delivery and birth status of the babies. Data analysis was done according to the statistical package for social sciences (SPSS) software version 12.0 and EpiInfo packages.

2.10. ETHICAL CONSIDERATION

This study was approved by the Kenyatta National Hospital–University of Nairobi and the Pumwani Maternity Hospital Ethics, Research and Standards Committees (KNH-UON ERC/01/3857 and PMH/DMOH/84/34, respectively). Informed consent was obtained from the mothers and confidentiality was ensured by use of in-patient numbers without names. Mothers with questions about anomalies were educated and anomalous babies were referred for management according to the laid down protocols of the respective hospitals.

2.11. STUDY BENEFITS

The benefits include my partial fulfilment of the requirements for the degree of master of dental surgery in oral and maxillofacial surgery; and it adds to the existing body of knowledge, serving as base-line material on CFAs for further research in Nairobi, Kenya. The information obtained will enable counselling of parents of children with CFAs using Kenyan specific data.

CHAPTER 3

RESULTS

The study covered an uninterrupted period of four and a half months. During this period there were 7989 new-borns manifesting 256 (3.2%) whole-body anomalies (anomalies in any part of the body). Out of the 7989 births, 4264(53.4%) were males, 3721(46.6%) were females and 4(0.05%) had ambiguous external genitalia. Males had more anomalies than females but the difference was not statistically significant.

KNH had 2410(30.2%) births, with 1262 males, 1145 females and 3 babies with ambiguous external genitalia. PMH had 5579(69.8%) births, 3002 males, 2576 females and one baby with ambiguous external genitalia. CFAs were 146, forming 57.0% of whole-body anomalies and 1.8% of total births (Table 1).

Table 1. Distribution of births by hospital, gender and birth status

PLACE	TOTAL BIRTHS	GENDER	FREQUENCY OF *ANOMALIES*	LIVE BIRTHS	STILL *BIRTHS*
PMH	5579	M – 3002 F – 2575 *AEG - 3*	141(*85)	5395(*44)	184(*41)
KNH	2410	M – 1262 F – 1145 AEG – 1	115(*61)	2228(*55)	182(*6)
TOTAL	**7989**	**M – 4264 F – 3721 AEG – 4**	**256(*146)**	**7623(*99)**	**366(*47)**

(*) Refers to CFAs; M-Males, F- Females, AEG- Ambiguous External Genitalia

Total whole-body (WB) anomalies in PMH were 141 (55.1%) and CFAs were 85(33.3%) of the total anomalies. Total whole-body anomalies in KNH were 115(44.9%) and CFAs were 61(2.5%) of the total anomalies. Total live-births were 7623 and they had whole-body malformations occurring at a rate of 2.4% (183 anomalous live-births), with 86(1.1%) males and 96(1.3%) live-birth females and 1 with ambiguous external genitalia having been affected. Stillbirths in the two hospitals were 366(4.6%), males were 176, females were 190; out of these 73(20%) stillbirths had whole-body anomalies with 29(7.9%) male and 13(3.6%) female stillbirths having the CFAs (Table 2). Though there were more female stillbirths, male babies were more anomalous than females. The occurrence of anomalies in the stillbirths was significantly higher than in the livebirths.

Table 2. Distribution of anomalies by hospital, gender and birth status n=256

PLACE OF BIRTH	BIRTH STATUS		GENDER		
	LIVE BIRTH	STILL BIRTH	MALE	FEMALE	AMBIGUOUS GENITALIA
KNH- WB	94 (36.7%)	21 (8.2%)	51 (19.9%)	47 (18.4%)	3 (1.2%)
CFA	55 (21.5%)	6 (2.3%)	37 (14.5%)	24 (9.4%)	
PMH- WB	88 (34.4%)	53 (20.7%)	84 (32.8%)	70 (27.3%)	1 (0.4%)
CFA	44 (17.2%)	41 (16%)	46 (18%)	39 (15.2%)	

Table 3 shows the age distribution of women in the study. The youngest was a 12-years-old primigravida while the oldest was a 47-

years-old who delivered her eleventh baby. The mean age of the mothers was 25.2 years; the mode was 24 years while the median age was 25years. The peak reproductive age range was 20-24 years. Women delivering in PMH were generally younger than those delivering at KNH. Major anomalies were most common in KNH. Most anomalies occurred during the peak reproductive age producing 88(34.4%) of the total anomalies. Extremes of ages (<15 and >35years) were not significantly associated with occurrence of anomalies (Table 4).

Table 3. Distribution of anomalies according to mothers' ages

CENTRE		AGE GROUP OF MOTHERS								
		NR	<15 years	15-19	20-24	25-29	30-34	35-39	40+	Statistical tests
Normal babies	KNH	495	1	162	673	770	400	146	33	X^2=188.16;6 df *p<0.01 (0.000)
	PMH	87	17	688	1966	1374	610	206	33	
Abnormal babies	KNH	6	0	8	40	29	18	1	0	X^2=13.63;5 df **p<0.05 (0.018)
	PMH	8	0	21	48	24	20	12	1	
TOTAL		596	18	879	2727	2197	1048	365	67	7897(GT*)

**p-significant differences at a=0.01: **p significant differences at a=0.05. n =7897(no. of mothers who delivered) due to the 52 twins delivered in KNH, 124 twins and 2 triplets delivered in PMH. GT* - Grand Total. NR (Not recorded) - Missing values or mothers who did not know their year of birth.*

In this study the peak birth weight was 3.0-3.9kg, peak anomalies, however, were noted in the 2.0-2.9Kg birth weight bracket. KNH had significantly more anomalous babies and a lower mean birth-weight (2.7Kg) than PMH (3.0Kg). The lowest baby-weight was 0.5kg and the highest 5.4 kg. CFAs were common at birth weight above 2.5Kg (60.2%) although weight below 2.5Kg. was significantly associated with whole-body anomalies (Tables 4 and 5).

Table 4 . Tests of significance for birth weight, ages of mothers and birth order in association with anomalies.

Variable	n per group		Odds	95%	P value
	Normal	Anomalous	ratio	confidence	
Birth weight					
<2.5kg	1399	113			
>2.5kg	5931	134	3.58	2.74 – 4.66	*0.000(P<0.01)
Birth order	3195	139			
1st borns	4423	106	1.82	1.40 – 2.37	*0.000(P<0.01)
others					
Mothers' age	6661	208			
<35 years	418	14	1.07	0.59 – 1.90	0.05 (0.912)
>35 years					

** Significantly associated with occurrence anomalies. NR values reduced the n per a class.*

Figure 1, shows the proportionality of anomalies according to birth weight which was done to rule out the thought that anomalies were more in a certain weight category just because more babies belonged to that weight category. Anomalies steeply decreased with increasing birth weight.

Table 5. Distribution of anomalies according to birth weight of babies

BABY	CENTRE	BIRTH WEIGHT OF BABIES							
		NR	**0.1-0.9kg**	**1.0-1.9kg**	**2.0-2.9kg**	**3.0-3.9kg**	**4.0kg and above**	**Mean weight & SD±**	Statistical tests
Normal	**KNH**	**204**	**71**	**196**	**663**	**1086**	**100**	**2.7 (±0.81)**	χ^2=207.09 ; 4df *p<0.01 (0.000)
	PMH	**217**	**23**	**210**	**1586**	**3200**	**178**	**3.04 (±0.548)**	
Abnormal	**KNH-WB**	**8**	**0**	**31**	**41**	**30**	**4**	**2.53(±0.82)**	χ^2=25.09; 4df *p<0.01 (0.000)
	PMH-WB	**0**	**18**	**17**	**58**	**36**	**12**	**2.52 (±0.98)**	
	OCFA KNH +PMH	**5**	**22**	**25**	**58**	**25**	**11**		
TOTAL		429	112	454	2348	4352	294	7989	

**p significant differences at a=0.01. WB- Whole body anomalies include OCFAs.*

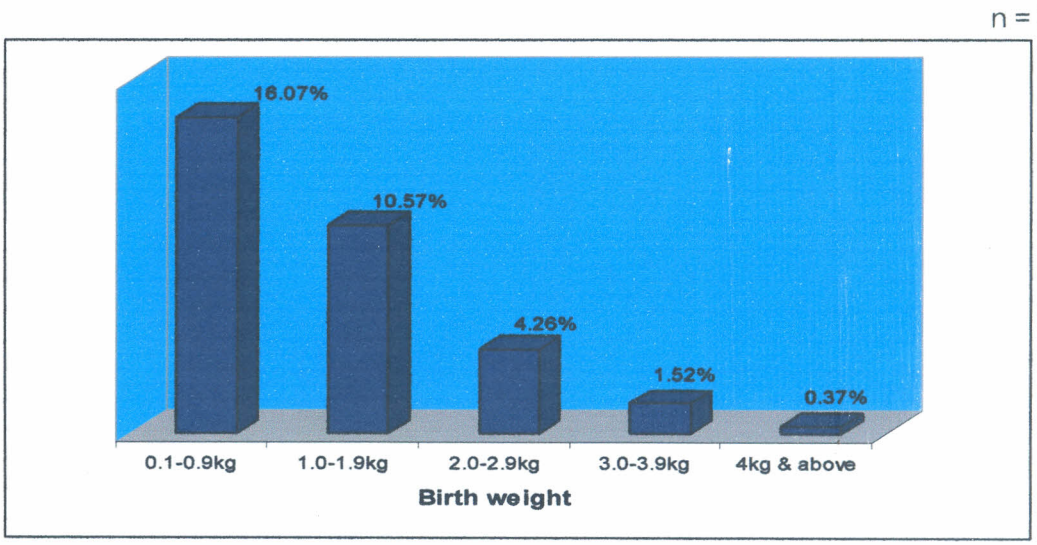

Fig. 1 Proportions of anomalies according to ranges of birth weight.

About 189(73.8%) of the babies born with anomalies were between the first and the second birth order (Table 6). There were no anomalies beyond 6th borns and to rule out the idea that the first borns seemed heavily laden with anomalies just because there were more first-borns in this study, a proportionality test was done which showed that sixth

borns had the highest proportion of anomalies, but the Fisher exact-test *(Fisher exact test x^2 =0.27: 1df p>0.05)* done to determine whether the 1st or 6th borns were the most predisposed confirmed that the first birth-order was significantly associated with anomalies (Fig. 2).

Table 6 Distribution of anomalies according to birth order

BABIES	*CENTRE*	*BIRTH ORDER*											
		NR	1	2	3	4	5	6	7	8	9	10	11	Total
Normal	KNH	38	803	696	416	194	82	36	17	7	1	1	2	2293
	PMH	49	2392	1555	798	371	151	54	23	12	2	1	4	5412
Abnormal	KNH	17	56	27	8	13	0	0	0	0	0	0	0	121
	PMH	13	83	23	19	11	0	5	0	0	0	0	0	154
TOTAL		117	3334	2301	1241	589	233	105	40	18	3	2	6	7989

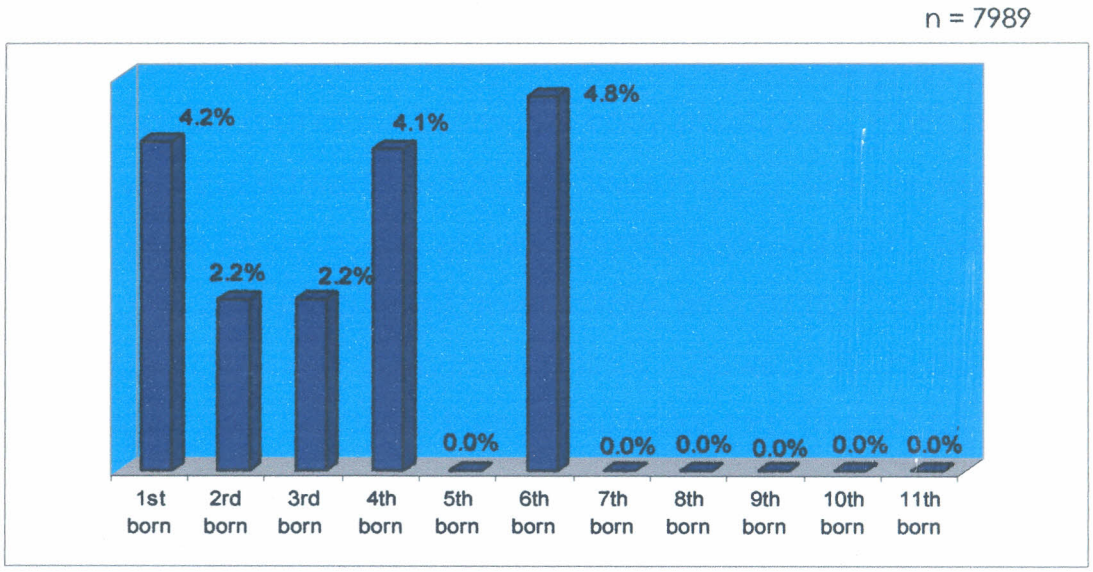

Fig.2. Proportions of anomalous babies within the birth-orders.

Out of the 5930 spontaneous vertex deliveries (SVDs) 148 (2.5%) had anomalies and out 2059 caesarean sections (CSs) 103 (5.0%) had anomalies. Anomalous babies were significantly delivered via CS

compared to the normal babies but there was no significant difference in the modes of delivery in the two hospitals (Table 7).

Table 7. Modes of delivery of anomalous babies in the two hospitals

Variable	Place of birth		Statistical test
	PMH	KNH	
Anomalous babies			
Mode of delivery			
SVD	89(1.1%)	60 (.8%)	
CS	52 (.7%)	55(.7%)	$\chi 2=3.12$: 1df:
Total	141(1.8%)	115(1.4%)	P>0.05(0.077)
Babies Without anomalies			
Mode of delivery	4345(54.4%)	1436(18%)	
SVD	1093(13.7%)	859((10.8%)	
CS	5438(68.1%)	2295(28.7%)	
Total			

The 146 CFAs were broadly classified into aural (65), orbital (15), cranial (35), oral (26) and isolated nasal anomalies (5) while the other body parts and systems were referred to according to the part that was involved (110 of the 256 whole body anomalies). When the whole body was considered preauricular sinus (Fig.3C) was the commonest anomaly at an incidence of 0.4% (4.3/1000) of total births, followed by extra digit at 0.3% (3/1000), then talipes at 0.2% (2.4/1000). The most common CFA was preauricular sinus followed by hydrocephalus (Fig.5A) at 0.2% (1.9/1000) then preauricular tags (Fig.3D) and cleft lip and palate (Fig.6A) at a prevalence of 1.5/1000 and 1.3/1000 respectively.

AURAL ANOMALIES

Aural anomalies (Table 8) formed 65(44.5%) of the 146 CFAs and occurred at a rate of 0.8% (8.1/1000) of the total births of which

preauricular sinus was the most common minor anomaly. It was also the most frequent whole body anomaly at 4.3/1000 births (0.4%). Microtia (Figs.3A,B) occurred at 0.1% of total-births; occurring in a spurt within one week ranging from the most severe (type III) to the least severe (type I). It was common in males and the right ear was the most involved. At birth, the position of the ears is generally lower in infants, but excessively low-set ears was observed in 6 infants giving a prevalence rate of 0.08% (0.8/1000 births). The syndromic low-set ear was noticed in 2 infants who had Down's syndrome (0.3/1000 births): this occurred in conjunction with posteriorly oriented ears, webbed neck and mongoloid slanting palpebral fissures. Imperforate external auditory meatus (Fig. 5E), preauricular cyst and atretic ears (thin ears) had the least incidence at 0.013% (0.13/1000 births). Preauricular sinus was common on the right, was single in all instances except in one case whereby two sinuses occurred with one superior to the other, both along the preauricular crease above the tragus. It occurred 1.5

times more in females than males. Preauricular tags (PTs) showed a preponderance to the right side with no gender predilection and both the sinuses and tags were observed more in live-births. PTs were mostly single and ranged from negligible skin elevations to large ones interfering with cosmetics. Some were the same colour as the surrounding skin while others were darker or lighter than the surrounding skin colour. PTs were noted to coexist with normal and abnormal helices at equal proportions. In one instance, three PTs of different colours and sizes were observed anterior to the auricle (Fig.3D).

Table 8. Distribution of aural anomalies by type

Table 8. Distribution of aural anomalies by type

AURAL ANOMALIES	FREQUENCY	% OF ANOMALY IN CLASS	% OF TOTAL BIRTHS	INCIDENCE / 1000
Preauricular sinus	34	54.8	0.43	**4.3**
Preauricular Tag	12	19.4	0.15	**1.5**
Microtia	6	9.7	0.08	**0.8**
Low-set ears	6	9.7	0.08	**0.8**
Imperforate Ex. auditory meatus	3	1.6	0.04	**0.4**
Atretic ears	2	1.6	0.03	**0.3**
Posteriorly oriented ears	1	1.6	0.01	**0.1**
Preauricular cyst	1	1.6	0.01	**0.1**
TOTAL	**65**	**100**	**0.81**	**8.1**

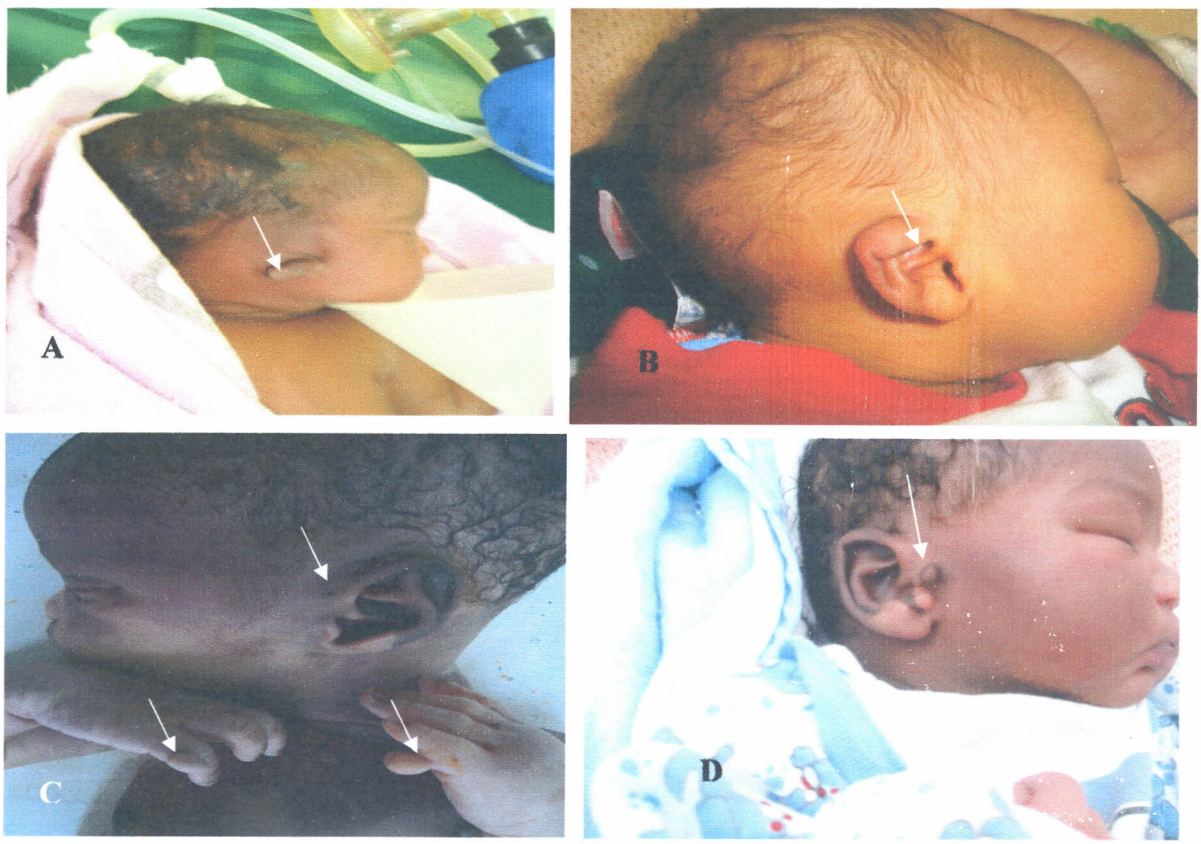

Fig.3. AURAL ANOMALIES: *A. Right Microtia II. B. Right Microtia I. C. Left Preauricular Sinus and bilateral ulnar polydactyly. D. Right Preauricular Tag*

ORBITAL ANOMALIES

Orbital anomalies were 15(1.9/1000) in 7989 births and formed 10.25% of CFAs. Microphthalmia (Fig.4B) occurred more commonly in orbital anomalies at rate of 0.05% (0.5/1000births), followed by hypertelorism (Fig.4B). Hypotelorism (Fig 4A), ankyloblepharon (Fig.4C), Mongoloid slanting palpebral fissures (Fig.4B), cyclopia (Fig.4D) and congenital glaucoma were noticed at 0.01% (0.1/1000 births).

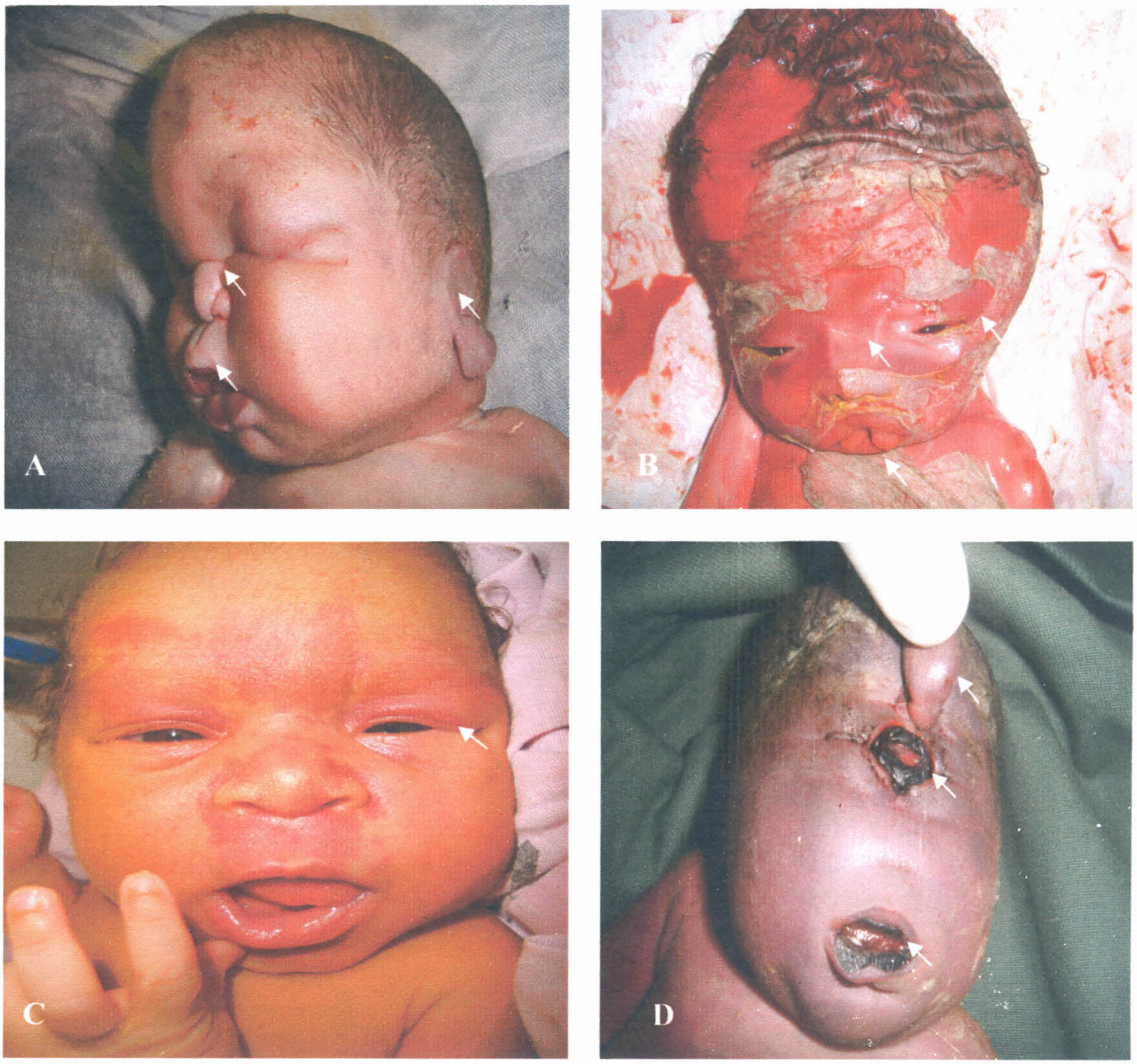

Figure 4: ORBITAL ANOMALIES: *A. Hypotelorism, depressed nasal bridge, adherent ears with imperforate external auditory meati, short upper lip. B. Microphthalmia, upward slanting palpebral fissures, wide depressed nasal bridge, glossoptosis, hypertelorism, C. Ankyloblepharon. D. Proboscis with cyclopia and microstomia.*

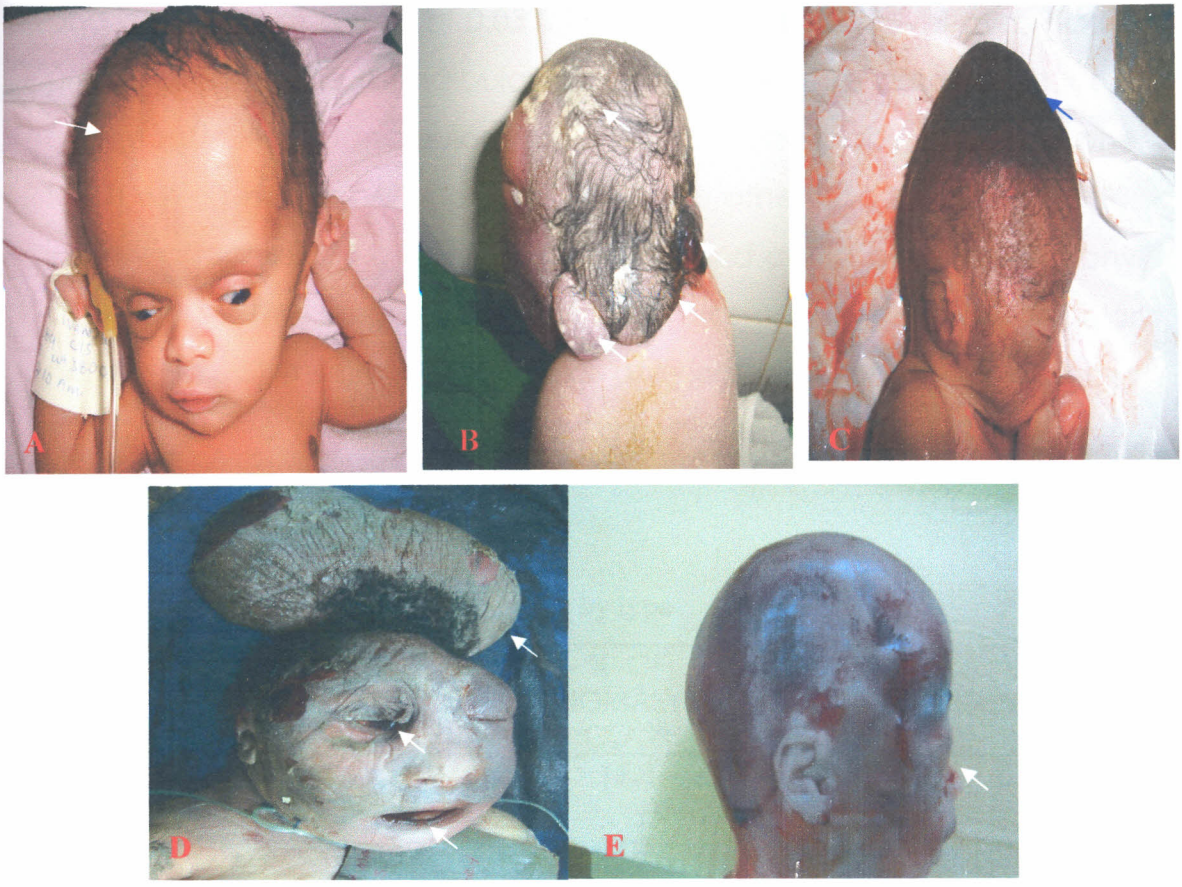

Figure 5. CRANIAL ANOMALIES: A. *Hydrocephalus B. Anencephaly, pseudencephaly, cup ears, head fused to thorax. C. Plagiocephaly. D. Triphyllocephally, macrostomia (the right eye was traumatized in the mortuary). E. Congenital alopecia, depressed nasal bridge and an imperforate external auditory meatus*

ORAL ANOMALIES

Oral anomalies were 26 out of 7989 births, occurring at a rate of 0.30% (3.0/1000 births) and formed 16.44% of CFAs (Table 10). Clefts of the lip and palate (Fig.6A) in combination formed the highest oral anomaly with a prevalence rate of 0.13% (1.3/1000 births). Clefts of the hard palate were the most common at 0.05% (0.5/1000 births) followed by high arched palate at a prevalence rate of 0.04% (0.4/1000 births). In about (7 out of the 10 clefts) three quarters of the cases, the clefting

was bilateral involving females and males equally. Gingival cysts (Fig.6B), natal teeth and micrognathia had no gender predilection and occurred at a rate of 0.03% (0.3/1000 births). Gingival cysts had a predilection for the mandibular posterior ridge while natal teeth were common in the mandibular incisor region. One baby had only one tooth which was firmly attached, while the other had two natal teeth, loosely attached by a soft tissue pedicle and could be moved in any direction by the tongue. Macrostomia, glossoschisis (Fig.4B), alveolar notch (Fig.6C) macroglosia, inferior ankyloglosia, congenital epulides and congenital ranula (Fig.6D) had a prevalence rate of 0.01% (0.1/1000 births)each.

Table10. Distribution of oral anomalies by type

ORAL ANOMALIES	FREQUENCY	% OF ANOMALY IN CLASS	% OF TOTAL BIRTHS	**INCIDENCE/1000**
Cleft alveolus	***4**	**15.4**	**0.05**	0.5
High arched palate	**3**	**11.5**	**0.04**	0.4
Gingiva cysts	**2**	**7.7**	**0.03**	0.3
Cleft lip	***2**	**7.7**	**0.03**	0.3
Cleft hard palate	***2**	**7.7**	**0.03**	0.3
Cleft soft palate	***2**	**7.7**	**0.03**	0.3
Natal teeth	**2**	**7.7**	**0.03**	0.3
Micrognathia	**2**	**7.7**	**0.03**	0.3
Macrostomia	**1**	**3.9**	**0.01**	0.1
Macroglossia	**1**	**3.9**	**0.01**	0.1
Inferior ankyloglossia	**1**	**3.9**	**0.01**	0.1
Congenital epuli	**1**	**3.9**	**0.01**	0.1
Congenital ranular	**1**	**3.9**	**0.01**	0.1
Alveolar notch	**1**	**3.9**	**0.01**	0.1
Bifid tongue	**1**	**3.9**	**0.05**	0.5
TOTAL	26	100.4	0.04	0.4

** Bilateral*

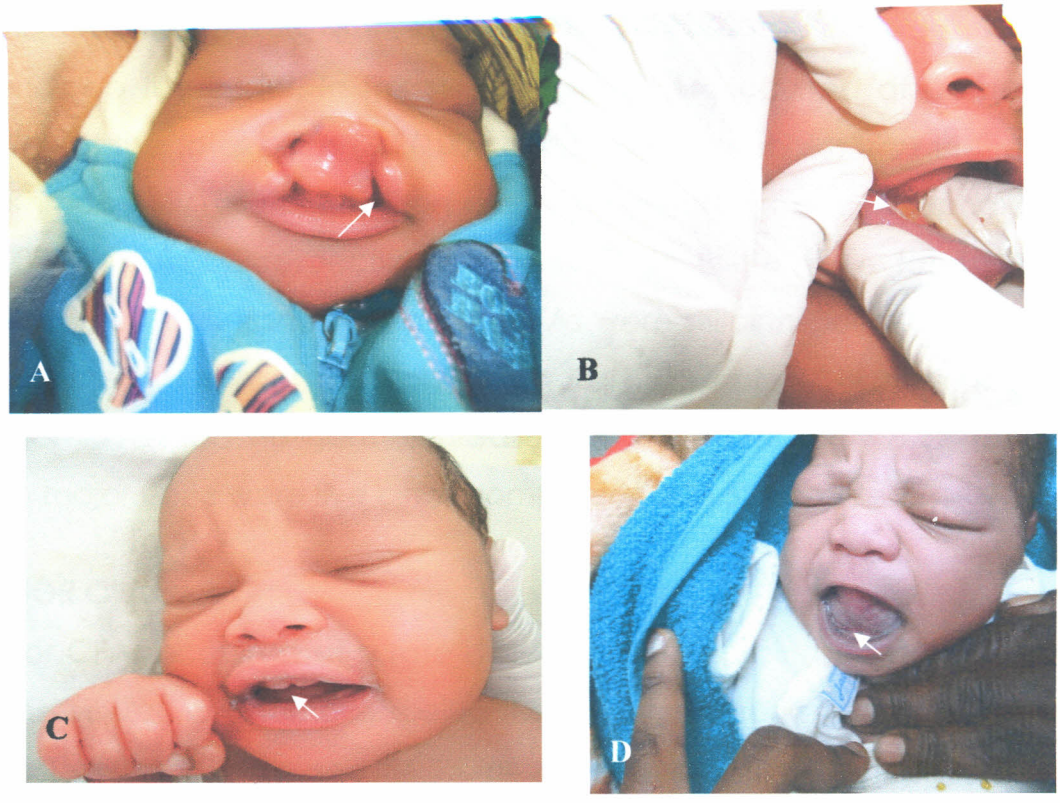

Figure. 6. ORAL ANOMALIES. *A. Bilateral cleft lip and palate. B. Gingival cyst. C. Alveolar notch. D. Congenital ranula.*

NASAL ANOMALIES

Isolated nasal (not associated with clefts) anomalies were rare, with a prevalence of 0.04% (0.4/1000 births). All of them occurred in association with other anomalies and all the cases were still-births. Depressed nasal bridge, depressed alae nasi and proboscis, each occurred at 0.01% (0.1/1000 births). The proboscis occurred together with cyclopia (Fig.4D) while the depressed nasal bridge and alae nasi occurred with severe scaphocephaly, atretic low-set ears, short neck and bilateral talipes.

MAJOR CFAs

Major (severe) congenital anomalies may be fatal or handicap the patient throughout life while minor anomalies such as skin tags do not handicap the infant. In this study major CFAs occurred at a rate of 0.61% of the total births (6.1/1000 births). Hydrocephalus was the most common (1.9/1000 births) then anencephaly and pseudencephaly at 0.08% and 0.06% respectively (Appendix III; Table 1). A total of 32 (65.30%) of the infants with major CFAs were stillbirths, and 42(85.71%) had other anomalies (multiple anomalies). The rate of occurrence of major CFAs had no gender predilection. However, only 8.2% males with major CFAs were live births compared to 26.5% females.

MINOR CFAs

Minor CFAs occurred at a rate of 1.1% (11.5/1000 births) among total-births with aural anomalies being the most common at 0.8% (8.0/1000 births) of total births and 66.0% of minor anomalies. Microphthalmia was the commonest minor orbital anomaly at 0.05% (0.5/1000) total births and forming 4.4% of the minor CFAs (Appendix III; Table 2). This was followed by high-arched palate at a prevalence rate of 0.04% (0.4/1000), and forming 3.3% of the minor CFAs. Hypertelorism, scaphocephaly, gingival cysts, natal teeth and micrognathia occurred at 0.03% (0.3/1000 births), while atretic ears, posteriorly oriented ears, hypotelorism, ankyloblepharon, mongoloid slanting palpebral fissure, failed closure of cranial sutures, trigonocephaly, plagiocephaly, turicephaly, clinocephaly, bulging fontanelle, macrostomia, macroglosia, inferior ankyloglosia, congenital epulides and preauricular cyst had a prevalence rate of 0.01% (0.1/1000) of total-births. Sixty (66.0%) of the minor CFAs were isolated cases but 31(34.8%) occurred with other anomalies. Of the ones with multiple anomalies 32.3% were associated with major anomalies and 67.1% with other minor anomalies.

CHAPTER 4

4.1. DISCUSSION

This study, just like those of Scheinfeld et al. (2004)[25] and Kohelet et al.(2002)[35] found preauricular sinus (PS) to have been the most common minor whole-body anomaly and the most common CFA at 4.3/1000 births. Preauricular sinus was common on the right, in females (F: M=1.5:1) and were either unilateral or bilateral. When unilateral they were commonly single and just anterior to the root of the helix. In the present study two preauricular sinuses were encountered on the right ear with one sinus above the other both along the preauricular crease above the tragus. Preauricular tags, just as reported by Durakbasa et al.[36] (2004) also showed preponderance to the right; they were more prevalent in males than females at 2:1 and bilateral in 50% of the cases. In one of the cases in this series there were bilateral multiple preauricular tags of different sizes and colours which could imply different soft tissue contents hence a varying origin.

Aketch (2000)[20] found an incidence of 1.9% of whole-body congenital abnormalities out of 7,125 babies and reported that the maternal age over 35 years and breech presentation were significantly associated with the birth of anomalous infants. Muga[37] (1985) in a study of 7,355 births at KNH found congenital anomalies at 2.8% of all births and cleft lip and palate at 1.1 per 1000 births. In the present series 7989 births occurred over four and a half months, manifesting 256 (3.2%) whole body anomalies. CFAs were 146(1.8%) of the total births. The lower rates of the anomalies (1.9%) reported by Aketch (2000) could have been because of the omission of the CFAs which were not reported in that study. The study by Muga (1985) reported on the major CFAs plus anomalies in other body systems hence the rise in the prevalence rate

to 2.8% of all births. Minor CFAs such as preauricular sinus, tags and gingival cyst were not reported. Evidently, the longer the duration of study the higher the sample size and the more the observers, the lower the prevalence rate. This could necessitate a modified study whereby the investigator is strictly stationed in one centre of delivery for a short period of time such as four weeks for 24 hours per day, allowing the examination of all the babies in details from head to toe hence eliminating inter-observer variation and fatigue. This could probably increase the prevalence rate of these anomalies and enable identification of various deformations and variations within normal.

As has been previously reported by various investigators,[8,25] microtia was common in males and on the right in unilateral cases. In bilateral cases the ears were either equal in size or different, with the right auricle being smaller or the most distorted than the left one and other aural anomalies such as preauricular sinus and imperforate external auditory meatus coexisted with microtia in about half of the cases. Non-syndromic microtia in this study occurred at a prevalence rate of 0.2/1000 live-births which concurred with the prevalence rate reported by Scheinfeld et al. [25] (2004). Khan et al.[3] (1977) in Lusaka, Zambia did a six-month study of single observations for congenital malformations on 8,505 children born at the University hospital and found an overall incidence of anomalies to have been 17.6 per 1000 births. Among the anomalous children 16.5 per 1000 live births had single malformations while multiple malformations occurred at 1.1 per 1000 neonates, with the highest incidences of these defects occurring in the first or second born neonates and in children born to females between 19-30 years. The birth order and maternal age most involved compared well with the present study but no classification of the anomalies into minor and

major was done, hence the difficulty in comparing his results with those of the present series.

Clefts of the lip and palate were the most common presenting in 10(1.3/1000) of the total births. Of these 9(90%) were bilateral cleft lip and palate and 1(10%) was left sided incomplete cleft lip. Only 3(30%) of these babies were live births with the other 7(70%) having been still-births. These concurred with the other reports in literature.[2,3,17] Half of the still-births with cleft lip and palate were also anencephalics in this series. Cleft lip and palate had a male to female proportion of 3:1 while the second most common oral anomaly in this study was the high arched palate occurring at 0.4/1000(3 of 7989) which was observed solely in females. Gingival cyst, natal teeth and micrognathia had no gender predilection and occurred at a rate of 0.03% (0.3/1000) of the total births. The prevalence of gingival cysts in this series was lower than the 11% reported by Dilley et al. but the prevalence of natal teeth (0.3/1000) was similar to that reported by Dilley et al.[28] Both cases of natal teeth were in the mandibular incisor region. The intraoral anomalies showed a low prevalence rate (0.01%) compared to other reports in literature [28,29] probably because they were missed due to the one-off examination within hours of delivery.

Just like in the previous studies which reported that congenital anomalies were common in the underweight (≤2.5kg),[14] this study also found that malformations occurred significantly in the underweight. Anomalies were not elicited below 14 years and were hardly observable above 45 years. The peak delivery age of 20-24 years compared well with other studies[11,12,38] but younger mothers gave birth to malformed babies in this study contrary to the study by Aketch (2000)[20].This series reports anomalous male: female ratio of 1.5:1, which

compares well with the previous reports[11,12,39] and about 74.1% of babies born with defects were in the first and second birth order just like in the previous studies.[3,11,12] The CS rate was much higher than the one reported by Wambua et al.[40] (1991) of 95% SVDs and 1.3% CSs.

Opinions differ regarding maternal age as a factor in the overall incidence of congenital defects. Hay etal.[11] (1972) and Chung etal.[12] (1975) found that congenital malformations tended to increase with advanced maternal age but this series and other workers in Kenya[37,39] did not find any significant difference in the effect of increased maternal age in the overall incidence of congenital malformations and Illesanmi et al.[41] (1998) in their Nigerian study of elderly primigravidae also did not find any association of anomalies with advanced maternal age. This may mean that African mothers who deliver babies with anomalies are relatively younger than non-Africans but it could also be due to the fact that African women give birth earlier. A multicentre series would be required to reach a definite conclusion.

Microphthalmia was the commonest orbital anomaly but orbital anomalies were generally rare occurring at less than 0.03%. Nasal aplasia with proboscis, in this series was only 1 (1.0/10,000) out of total births, producing a higher incidence in this series than the one reported by Moore and Persand (2003)[14] at 1:100,000 newborns. Cranial anomalies occurred at 0.4% (4.4/1000 births) and constituted 23.3% of CFAs. Hydrocephalus was the most prevalent at 15(1.9/1000) in the 7989 total births, giving twice the prevalence of 0.4-0.8/1000 total births reported by other investigators.[8,25] Anencephaly which was the second commonest occurred at 0.08% (0.8/1000) births; it had no gender predilection and was incompatible with life.

The incidence of congenital malformations in live births in a given population tends to increase with follow-up period after birth. McIntosh etal.[5] could only diagnose 43.2% of all malformations at birth but at the end of a 1-year follow-up, the figure had gone up to 97%. There was no follow-up in this study and some of the malformations could most likely have been missed due to the one-off examination. In Kenya, these anomalies are currently managed by local and visiting plastic surgeons, oral and maxillofacial surgeons, ear nose and throat surgeons and the general surgeons in almost any hospital with a theatre. There is no laid down protocol of management of these cases and no follow up especially for the patients operated on during the free surgical camps. The management of CFAs is usually multidisciplinary involving at least genetic counsellors, paediatricians, plastic surgeons, oral and maxillofacial surgeons, specialist nurses, ear nose and throat surgeons, orthodontists, speech therapists and prosthodontists who would work very well in specific CFA centres for the benefit of the nation, clinicians, parents and the babies.

4.2. CONCLUSIONS

The pattern obtained in this study revealed that a younger age group of mothers delivered infants with malformations with the commonest CFAs being preauricular sinus, hydrocephalus, clefts of the lip and palate. These anomalies were common in the first and second birth-order, particularly in babies delivered via caesarean section and among stillbirths. Male and low birth weight babies were the most involved.

4.3. LIMITATIONS OF THE STUDY

1. Kenyan women of low socio-economic status hardly deliver in hospitals and are more likely to be delivered in hospitals if they have obstetric problems, which malformations sometimes cause.[8]
2. In-breeding may precipitate an increased rate of malformations. However, the communities (Arabs, Asians, and Somalis) who practise this culture hardly visit the study sites.
3. The mothers were unwilling to talk when in labour or immediately after delivery due to pain and exhaustion.
4. Due to the low staff to patient ratio, the forms were sometimes filled in hurry leading to some omissions during entry hence (NR) missing data and erroneous entries. Other patients did not know their years of birth while breakdown of weighing machine in PMH led to a high number of unrecorded weights of neonates.
5. The intraoral anomalies showed a low prevalence rate (0.01%) compared to other reports in literature[28,29] probably because they were missed due to the one-off examination within hours of delivery.

4.4. RECOMMENDATIONS

1. The CFAs are evidently rare, but Kenya does not have a protocol or a centre for their management hence the need to encourage health services to be organised so that specified centres treat more patients with CFAs allowing expertise to develop and the effectiveness of various treatment modalities to be evaluated objectively.

2. The study illustrated that younger mothers are delivering babies with major anomalies and that the anomalies may manifest in spurts. This could point to an existence of a terratogen at a particular time of gestation necessitating a study on the possible environmental aetiologic factors.
3. The information obtained enables counselling of parents of children with CFAs using Kenyan specific data. Folic acid and vitamin supplements reduce the incidences of neural tube defects and all mothers should be encouraged to take them prenatally.

REFERENCES

1. Stricker M, Van der Meulen J G, Raphael B, Tolhurst D E, Murray E J(Ed). Craniofacial malformations. Edinburg: Churchill Livingstone. 1990. pp149-309.
2. Chaurasia's Human Anatomy: Head, Neck and Brain. 3rd Ed. India: CBS Publishers and Distributors. 2003. pp309-313.
3. Khan A A, Ivanov I. Congenital malformations in Zambian Neonates. East African Med. J. 1977; 54:73-77
4. Russell R C G, William N S, Bulstrode C J K (Editors): Bailey and Loves' Short Practice of Surgery: Cleft Lip and Palate – Developmental Abnormalities of the Face, Mouth and Jaws. 24th Ed. London. Arnold Publishers. 2004. pp645-663.
5. McIntosh R, Katharine M K, Mary R R, Mary H S, Bellows T M. The incidence of congenital malformations. A study of 5,964 pregnancies. Paediatr J.1954; 14:505-507.
6. O'Doherty N. Congenital abnormalities; Atlas of the Newborn 5. London; Waterlow (Dunstable) LTD. 1975.pp1-95.
7. Moore L K, Persand T V N. The developing Human: Clinically Oriented Embryology, 7th Ed. Philadelphia; Elsevier Science.2003.pp158-480.
8. Kalter H, Warkany J. Congenital malformations: Aetiological factors and their role in prevention (first of two parts). The New England J. of Med. 1983; 308: 424-431.
9. Deuden B I, Reid T M S, Jewsbury J M, Turk D C. A new short Textbook of Microbial and Parasitic Infections. London: Butler and Tunner Ltd. 1988.pp320-329.
10. Day W D. Genetics of Congenital lip defects; Clinics in plastic surgery. Philadelphia. W.B. Saunders Co. 1984; 11:693-700.

11. Hay S, Barbano H. Independent effects of maternal age and birth order on the incidence of selected congenital malformations. Teratol J.1972; 6:271-279.

12. Chung C S, Myrianthopoulos N C. Factors affecting risks of congenital malformations: Analysis of epidemiologic factors in congenital malformations. Report from the collaborative perinatal project. Birth Defects Orig. Art. Series.1975; 11:1-22.

13. Yadav N S. Short Questions and Answers in Oral medicine and Oral pathology: Developmental Disorders. New Delhi; ATTBS Publishers and Distributors. 2001. pp41-75.

14. Murray J C, Daack-Hirsh S, Buetow K H, Mungeu R, Espina L, Paglinawan N, Villanueva E, Rary J, Magee K, Magee W. Clinical and epidemiologic studies of cleft lip and palate in the Philippines. Cleft Palate Craniofac J. 1997; 34:7-10.

15. Al-Omari F, Al-Omari I.K. Cleft lip and palate in Jordan: Birth prevalence rate. Cleft Palate Craniofac J. 2004; 41:609-612.

16. Iregbulem L M. The incidence of cleft lip and palate in Nigeria. Cleft Palate J 1982; 19:201-205.

17. Jain B R, Krogan M W. Cleft type and Sex differences in Craniofacial growth and Clefting from one month to ten years. Cleft Palate J. 1983; 20:238-245.

18. Fasana F. Textbook of Medical Embryology: Normal and Abnormal development. Nairobi; KLB. 1980. pp50-184.

19. A Non-Governmental Organization of WHO (World health organization). Congenital Malformations Worldwide: A report from the International Clearing House of Birth Defects Monitoring Systems. Amsterdam; Elsevier Science publisher Co. Inc.1991. pp 9-177.

20. Aketch O M. Profile of mothers who gave birth to babies with congenital anomalies in KNH. MMed. (Obs and Gyn.) Thesis, University of Nairobi, 2000.

21. Gorlin R J, Cohen M M, Hennekam R C M: Syndromes of the Head and Neck. 4th Ed. Oxford. Oxford University Press. 2001. pp379-972.

22. Screan J ST and Connor C. Congenital mucoceles: Report of two cases. Int. J. of Paed. Dentistry.1996; 6:271-275.

23. Persaud T V N(Ed). Abnormal embryogenesis: Cellular and molecular aspects. Baltimore, Maryland; University Park press. 1979. pp171-176.

24. Avery K J: Essentials of Oral Histology and Embryology: A Clinical Approach. 2nd Ed. Missouri. Mosby Inc. 2000. pp 36-50.

25. Scheinfeld SN, Silverbeg BN, Weinberg MJ, Nazad V: The Preauricular Sinus: A Review of its Clinical Presentation, Treatment and Associations. Pediatric Dermatology J.2004; 21:191-196.

26. Wilson G J, Clarke F F (Editors). Hand book of Teratology: Comparative Maternal and Epidemiologic Aspects. New York; Plenum Press. 1977. Pp244.

27. Tomizawa M, Kohno M, Noda T, Suzuki M. Three cases of Palatal Polyps in Infants. Int. J. of Paed. Dentistry1999; 9:213-217.

28. Dilley C D, Siegel A M, Budnick S. Diagnosing and treating common Oral pathologies: Paediatric clinic of North America 1991, 38.pp110-115.

29. Hunter M L, Roberts G J: Oral and Dental Anomalies in Ellis Van Creveld Syndrome. Int. J. of Paediatric Dent. 1988; 8: 153-157.

30. Cawson R A. Essentials of Dental surgery and pathology: Oral Diseases in the Children. Singapore; ELBS imprint. 1991; pp27, 511.

31. Okoth-Olende C A. Analysis of Congenital Choanal Atresia as seen at the KNH, NBI. 1974-1988. MMed (ENT) Thesis. University of Nairobi.1990.

32. Yang J, Carmichael S L, Kaidarova Z, Show GM. Risks of Selected Congenital Malformations among Offspring of Mixed Race-ethnicity. Birth defects Res. A. Clin Mol. Teratol. 2004; 70:820-824.

33. Wang Y R. Syndromic Ear Anomalies and Renal Ultrasounds. Pediatrics J 2001; 108: 32-33.

34. Nyaga W R. A Ten Year Retrospective study on Encephalocoeles as seen and managed at KNH. Jan.1992 - Dec2001. MMed (Surg.) Thesis University of Nairobi. 2003.

35. Kohelet D, Arbel E: A Prospective Research for Urinary Tract Abnormalities in Infants with isolated Preauricular tags. J. Pediatrics 2002; 141:388-391.

36. Durakbasa UC, Mutus M, Meric F, Sirlioglu E, Ayhan Y, Dedeoglu S, Sehiralti V, Tosyali N, Audac N. Prevalence of Preauricular Tags and Renal anomalies. Marmar Medical J. 2004; 17:109-112.

37. Muga O R. The incidence of Congenital Malformations in Newborns at KNH, Nairobi. MMed. Thesis. University of Nairobi. 1985.

38. Tadesse E, Yusuf L, Zein A Z. Retrospective Cross-sectional Forms of Deliveries at Gondar College of Medical Science (GCMS) from January 1985 to 1989. J.Obst. Gyn. East. Cent. Africa 1992; 10:77-79.

39. Fletcher M A. Physical Diagnosis in Neonatology. Philadelphia: Lippincott-Raven.1988. pp. 142-229.

40. Wambua L, Nasah B T, Leke R J I Ojwang S B O. Determinants of Perinatal Mortality: Hour and Day of birth, and Method of Delivery at the Central maternity, Yaounde. J. Obst. Gyn. East. Cent. Africa 1991; 9:72-73.

41. Illesanmi A O, Fawole O, Olaleye D O, Arowojolu A: Pregnancy Outcome in the Elderly Primigravidae in Nigeria. J. Obst. Gyn. East. Cent. Africa 1998; 14: 17-20.

APPENDIX I : Interview and Examination Form

CRANIOFACIAL ANOMALIES (CFAs) AMONG HOSPITAL BIRTHS AT KNH AND PMH, IN NAIROBI

I. Introduction

This study intends to determine the occurrence and distribution CFAs in the Nairobi births.

Ladies admitted to labour ward and maternity departments for delivery in Pumwani and Kenyatta Hospitals are requested to consent for an interview and a detailed examination to be done on their babies within 48 hours.

II. Personal Information (Tick or write as appropriate)

Mother's IP number.....

Centre code KNH [1] PMH [2]

1. Age of mother (years) .....
2. Birth order of the child.....
3. Number of previous live-births.....
4. Baby IP Number.....
5. Centre : KNH [1] PMH [2]
6. Baby: Live Infant [1] Stillbirth [2]
7. Gender: Male [1] Female [2]
8. Birth Weight ..... kilograms
9. Mode of Delivery
10. Caesarean [1] Vaginal (SVD) [2]
11. Presentation: Cephalic [1] Breech [2]

III. Anomalies

1. Presence of any anomaly? Yes [1] No [2]

2. Anomalous part
   i. Cranial
   ii. Ocular
   iii. Oral
   iv. Nasal
   v. Ear
   vi. Vascular /Cutaneous
   vii. Others

3. Magnitude
   i. Major: (a) Single (b) Multiple
   ii. Minor: (a) Single (b) Multiple

4. Types of Anomalies

i. OROFACIAL

1. Cleft lip	(a) Minimal (b) Full length	-Unilateral – • Not involving alveolar ridge • Involving alveolar ridge • Involving ridge and palate
2. Naso – ocular cleft		
3. Cleft palate	(a) Bifid uvula (b) Soft palate only	(c) Hard palate only (d) both soft and hard palate
4. Maxillomandibular (macrostomia) cleft	(a) Soft tissue only	(b) Soft tissue and bone

5. Combined cleft lip and palate defects	(a) Unilateral Complete (b) Unilateral incomplete	(c)Complete bilateral cleft lip and palate (d) Incomplete bilateral cleft lip and palate (e) severe cleft lip (f) Midline maxillary cleft (hare lip)
6. Mandibular anomalies:	(a) Coloboma (b) Bilateral cleft	(c) Midline mandibular cleft (d) Pits/fistulae
7. Tongue anomalies:	(a) Split tongue (b) Lobulated tongue (c) Accessory tongue (d) Macroglossia (e) Aglossia	(f) Scrotal tongue (g) Ankylotic tongue (h) Macroglosia (i) Lingual cyst/Fistulae
8. Jaw anomalies:	(a) Macrognathia (b) Ageniocephaly (c) Agnathia (d) Microstomia	(g) Micrognathia (h) Astomia (i) Hemifacial microsomia
9. Oral Mucosa	(a) Leukoedema	(a) White spongy naevus
10. Salivary Glands	(a) Ranular	(b) congenital mucoceles
11. Gingival and Alveolar anomalies	(a) Neonatal epuli (b) Alveolar lymphangiomas (c) Congenital fusion of gums (unilateral/ Bilateral)	(d)Idiopathic gingival fibromatosis (e) Eruption cyst (f) Alveolar cyst
12. Dental anomalies	(a) Natal teeth	
13. Other Congenital lip anomalies	(a) Thick or thin lips(double lip) (b) Aberrantly attached frenula (c) Short commissural length	(d) Pigmented naevi (e) Tissue mounds (f) Short/Long upper or lower lip

14. Nerve anomalies	(a) Facial nerve palsy	(b) Neurofibromatosis
15. Mimicry muscles	(a) Macrosomic infant (b) Absence of mimetic muscle (Crying face syndrome)	(c) Facial hemihypertrophy (d) Fibrous circumoral band(whistling face deformity)
16. Vascular anomalies	(a) Haemangioma	(b) Lymphangioma
17. Cutaneous anomalies	(a) Ectodermal dysplasia (b)Circumoral and oral black pigmentation	(c) Skin defects (absence) (d) congenital alopecia (e) Hypopigmentation of skin

ii. EAR (AURAL)

1. Adherent 2. Anotia	3. cysts 4. Microtia	5. Prominent ears 6. Preauricular sinuses	7. Tags 8. Others

iii. NASAL

1. Arhinia 2. Broad nose/flat bridge 3. Choanal atresia 4. Hypoplastic nose	5. Nasal aplasia 6. Nasoschizis 7. Nasal duplication 8. Nasal aplasia with proboscis	9. Small upturned nose

iv. CRANIAL

1. Acrania 2. Hydrocephalus 3. Microcephaly	4. Encephalocoele (a) Posterior (b) Anterior 5. Macrocephaly

v. OCCULAR

1. Anophthalmia 2. Anorbitism 3. Ankyloblepharon 4. Blue sclera 5. Blepharoptosis 6. Blepharoschizis 7. Blepharophimosis	8. Cebocephaly 9. Cryoptophthalmia 10. Cyclopia 11. Congenital aphakia 12. Canthoschizis 13. Canthal dystopia 14. Epicanthus 15. Euryblepharon	16. Epiblepharon 17. Ethmocephaly 18. Microphthalmia 19. Microblepharon 20. Synophthalmia 21. Telecanthus 22. Others: Specify...

OTHERS:
SPECIFY.....
.....
.....
.....

APPENDIX II
CRANIOFACIAL ANOMALIES AMONG BIRTHS AT KNH AND PMH IN NAIROBI

CONSENT FORM

Dear Parent/Guardian

I am a postgraduate student at the University of Nairobi pursuing studies specializing in Head and Neck surgery. I wish to request for your permission for you and your child to participate in a study that will form part of my degree work. The aim is to document the incidence and distribution of mouth, head and neck anomalies clinically manifest at birth in Pumwani Maternity and Kenyatta National Hospitals in Nairobi. From this study it is hoped that there will be improvement in the assessment and management of infants and provision of accurate information to famiiies.

The study will involve answering to some questions by you and a detailed examination of your child. which will be done by the investigating team. I also request that you allow photography of any anomaly seen. All the findings will be recorded and later analyzed for this research purpose only. Should any condition be detected in your child, they will be referred for specialist attention or you will be advised accordingly. No invasive procedure will be performed on your child during the study and no extra costs will be caused by the study to you.

I would therefore appreciate your consent by signing below.

I, ... Parent/Guardian of .....

P.O BOX.....

Do hereby freely consent / do not consent to my child and me to participate in the current oral and craniofacial study.

Dr/Mr/miss/Mrs ..... has explained what is required of my child and me. I understand that this consent will not alter any planned medical or/surgical care to me or my child. I am also informed and understand that all information about us shall be treated in the strictest confidence.

Signed..... **Date**.....
(Parent or guardian)

Witnessed by..... **Date**.....
Signature

APPENDIX III

Table 1: Distribution of Major CFAs

MAJOR CFAs	FREQUENCY	% OF ANOMALY IN CLASS	% OF TOTAL BIRTHS	**INCIDENCE /1000**
Hydrocephalus	**15**	**31.2**	**0.19**	1.9
Anencephaly	**6**	**12.5**	**0.08**	0.8
Encephalocoele	**5**	**10.4**	**0.06**	0.6
Cleft hard palate	**4**	**8.3**	**0.05**	0.5
Cleft lip	**2**	**4.2**	**0.03**	0.3
Cleft alveolar	**2**	**4.2**	**0.03**	0.3
Cleft soft palate	**2**	**4.2**	**0.03**	0.3
Depressed nasal bridge / alae (x2)	**2**	**4.2**	**0.03**	0.3
Absence of eyelashes	**2**	**4.2**	**0.03**	0.3
Absence of eyebrows	**2**	**4.2**	**0.03**	0.3
Imperforate external auditory meatus	**1**	**2.1**	**0.01**	0.1
Cyclopia	**1**	**2.1**	**0.01**	0.1
Congenital glaucoma	**1**	**2.1**	**0.01**	0.1
Congenital ranula	**1**	**2.1**	**0.01**	0.1
Proboscis	**1**	**2.1**	**0.01**	0.1
Congenital alopecia	**1**	**2.1**	**0.01**	0.1
TOTAL	48	100	0.08	0.8

Table 2: Distribution of Minor CFAs.

MINOR CFAs	FREQUENCY	% OF ANOMALY IN CLASS	% OF ALL BIRTHS	**INCIDENCE /1000**
Preauricular sinus	**34**	**36.96**	**0.43**	4.3
Preauricular tag	**12**	**13.0**	**0.15**	1.5
Microtia (anotia)	**6**	**6.5**	**0.08**	0.8
Low-set ears	**6**	**6.5**	**0.08**	0.8
Microphthalmia	**4**	**4.4**	**0.05**	0.5
High arched palate	**3**	**3.3**	**0.04**	0.4
Hypertelorism	**2**	**2.2**	**0.03**	0.3
Scaphocephaly	**2**	**2.2**	**0.03**	0.3
Natal teeth	**2**	**2.2**	**0.03**	0.3
Micrognathia	**2**	**2.2**	**0.03**	0.3
Atretic ears	**1**	**1.1**	**0.01**	0.1
Posteriorly oriented ears	**1**	**1.1**	**0.01**	0.1
Hypotelorism	**1**	**1.1**	**0.01**	0.1
Ankyloblepharon	**1**	**1.1**	**0.01**	0.1
Mongoloid slanting palpebral fissures	**1**	**1.1**	**0.01**	0.1
Trigonocephaly	**1**	**1.1**	**0.01**	0.1
Wide open sutures	**1**	**1.1**	**0.01**	0.1
Plagiocephaly	**1**	**1.1**	**0.01**	0.1
Turicephaly	**1**	**1.1**	**0.01**	0.1
Clinocephaly	**1**	**1.1**	**0.01**	0.1
Bulging fontanelle	**1**	**1.1**	**0.01**	0.1
Gingival cysts	**1**	**1.1**	**0.01**	0.1
Macrostomia	**1**	**1.1**	**0.01**	0.1
Macroglosia	**1**	**1.1**	**0.01**	0.1
Inferior ankyloglosia	**1**	**1.1**	**0.01**	0.1
Congenital epuli	**1**	**1.1**	**0.01**	0.1
Alveolar notch	**1**	**1.1**	**0.01**	0.1
Bifid tongue	**1**	**1.1**	**0.01**	0.1
Preauricular cysts	**1**	**1.1**	**0.01**	0.1
TOTAL	92	100.4	1.15	11.5

Table 3. Distribution of anomalies in other parts of the body

SYSTEM	FREQUENCY	% OF ANOMALY IN CLASS	% OF ALL BIRTHS	**INCIDENCE /1000**
Urogenital System				
Ambiguous external genitalia	**4**	**33.3**	**0.05**	0.5
Undescended testis	**2**	**16.7**	**0.03**	0.3
Phimosis	**2**	**16.7**	**0.03**	0.3
Scrotal hernia	**1**	**8.3**	**0.01**	0.1
Coiled penis	**1**	**8.3**	**0.01**	0.1
Hypospadias	**1**	**8.3**	**0.01**	0.1
Hyperspadias	**1**	**8.3**	**0.01**	0.1
Subtotal	12	100	0.15	**1.5**
Appendicular anomalies				
Extra digits	**22**	**40.7**	**0.28**	2.8
Talipes	**20**	**37.1**	**0.25**	2.5
Everted lower limb (plantar version)	**3**	**5.6**	**0.04**	0.4
Over-ridding toes	**2**	**3.7**	**0.03**	0.3
Congenital digital amputation	**1**	**1.9**	**0.01**	0.1
Lower limb hypoplasia	**1**	**1.9**	**0.01**	0.1
Flexion deformity of the knees	**1**	**1.9**	**0.01**	0.1
Clubbed hand	**1**	**1.9**	**0.01**	0.1
Extra limb (5 full limbs)	**1**	**1.9**	**0.01**	0.1
Hypodactyly (thumb agenesis)	**1**	**1.9**	**0.01**	0.1
Vestigial limbs / digits	**1**	**1.9**	**0.01**	0.1
Subtotal	54	100	0.68	**6.8**
Spinal Dysraphism and Vertebral Anomalies				
Spina bifida	**14**	**77.8**	**0.18**	1.8
Failed closure of vertebral column	**2**	**11.1**	**0.03**	0.3
Lumbar kyphosis	**1**	**5.6**	**0.01**	0.1
Congenital scoliosis	**1**	**5.6**	**0.01**	0.1
Subtotal	18	100	0.23	**2.3**
Others				
Multiple anomalies (unclassified)	**6**	**23**	**0.08**	0.8
Gastroschisis	**4**	**15.4**	**0.05**	0.5
Anal tag	**4**	**15.4**	**0.05**	0.5
Umbilical hernia	**4**	**15.4**	**0.05**	0.5
Short neck	**2**	**7.6**	**0.02**	0.3
Imperforate anus	**2**	**7.6**	**0.02**	0.3
Distended flabby abdomen	**1**	**3.8**	**0.01**	0.1
Webbed neck	**1**	**3.8**	**0.01**	0.1
Extrocardia	**1**	**3.8**	**0.01**	0.1
Omphalocoele	**1**	**3.8**	**0.01**	0.1
Subtotal	**26**	**100**	**0.31**	**3.1**

KENYATTA NATIONAL HOSPITAL
Hospital Rd. along, Ngong Rd.
P.O. Box 20723, Nairobi.
Tel: 726300-9
Fax: 725272
Telegrams: "MEDSUP", Nairobi.
Email: KNHplan@Ken.Healthnet.org

Ref: KNH-ERC/ 01/ 3857
Date: 25th October, 2006

Dr. Odhiambo Atanasias,
Dept. of Oral and Maxillofacial Surgery,
Faculty of Dental Sciences,
University of Nairobi.

Dear Dr. Odhiambo,

RESEARCH PROPOSAL: "RANGE AND PATTERN OF OCCURRENCE OF CLINICALLY MANIFEST CONGENITAL ORAL AND CRANIOFACIAL ANOMALIES AMONG BABIES BORN AT KENYATTA NATIONAL HOSPITAL AND PUMUANI MATERNITY HOSPITAL IN NAIROBI " (P170/8/2006)

This is to inform you that the Kenyatta National Hospital Ethics and Research Committee has reviewed and approved your above cited research proposal for the period 25th October, 2006 – 24th October, 2007.

You will be required to request for a renewal of the approval if you intend to continue with the study beyond the deadline given.

On behalf of the Committee, I wish you fruitful research and look forward to receiving a summary of the research findings upon completion of the study.

This information will form part of database that will be consulted in future when processing related research study so as to minimize chances of study duplication.

Yours sincerely

PROF A N GUANTAI
SECRETARY, KNH-ERC

c.c. Prof. K. M. Bhatt, Chairperson, KNH-ERC
The Deputy Director CS, KNH
The Dean, Faculty of Dental Sciences, UON
The HOD, Medical Records, KNH
Supervisors: Dr. Chindia, School of Dental Sciences, UON
Dr. M Ndavi, Dept of Obs & Gynae, UON
Dr. F. Macigo, School of Dental Sciences, UON
Dr. F. Were, Dept. of Paediatrics & Child Health, UON

PUMWANI MATERNITY HOSPITAL

Tel: 02/6763291– 4
Fax: 02/6762965

P.O. Box 42849
Code: 00100- GPO
Nairobi.

PMH/DMOH/84/34

2ND NOVEMBER 2006

To:
Dr. Odhiambo Atanasias

RE: RESEARCH PROPOSAL

The hospital Research and Ethics refers to the proposal you presented to us titled: *"Range and pattern of congenital oral and craniofacial anomalies clinically manifest at birth in Kenyatta National Hospital and Pumwani Maternity Hospital in Nairobi"*.

The committee has no objection and thus you can commence the study with adherence to hospital regulations and thereafter submit a copy of the findings to the hospital.

DEPUTY MEDICAL OFFICER OF HEALTH
PUMWANI MATERNITY HOSPITAL

DR C. WANYONYI
AG. MEDICAL SUPERINTENDENT

.c. File