Spectrum of congenital anomalies among newborns from selected Sub-Saharan African tertiary hospitals: focus on Zambia

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Presentation Focus and Question

- Review of Published Literature on Clinical and Social Demographic Characteristics of Congenital Anomalies among Newborns Presenting at Selected SubSaharan Tertiary Health Facilities with a Focus on Zambia

- What is the Plight of a Patient with Rare Disease in Africa (Sub-Saharan Africa)?

Disclosures

None

Key References

- Christianson A, et al (2006). Global Report on Birth Defects-the Hidden Toll of Dying and Disabled Children. March of Dimes Birth Defects Foundation, White Plains, New York **2006.**

- Uwineza A, et al. Array-CGH analysis in Rwandan patients presenting development delay/intellectual disability with multiple congenital anomalies. BMC Medical Genetics **2014;** 15:79

- Kishimba RS, et al. Factors associated with major structural birth defects among newborns delivered at Muhimbili National Hospital and Municipal Hospitals in Dar Es Salaam, Tanzania 2011– 2012.Pan African Medical Journal **2014;** 19:85 doi:10.11604/pamj.2014.19.85.3428

- Kalisya LM., et al. Patterns of Congenital Malformations and Barriers to Care in Eastern Democratic Republic of Congo. PLoS ONE10(7) **2015**; e0132362. doi:10.1371/journal.pone.0132362

- Mekonen HK, et al. Birth weight by gestational age and congenital malformations in Northern Ethiopia. BMC Pregnancy and Childbirth **2015,** 15:76

Definition

A birth defect is an abnormality of structure or function which originates during intrauterine development and is evident before birth, at birth or manifests later in life.

(Christianson RE et al., 1981; WHO)

Background: Global Burden of Birth Defects

(Christianson A, et al (2006). Global Report on Birth Defects-the Hidden Toll of Dying and Disabled Children. March of Dimes Birth Defects Foundation, White Plains, New York **2006.)**

- 7.9 Million are born each day and 6% of these with birth Defects
- 3.3million under-five year olds annually of birth defects
- 3.2 of those that survive have life –long disability
- Globally 94% of severe birth defects burden and 95% of mortality are in low-middle income countries
- Prevalence of Birth Defects is variable Globally ranging from 39.7-82/1000 live births
- Top 5 are: 1)CHD, 2)Neural tube Defects,3)Haemoglobinopathies,4) Down Syndrome and 5)G6PD

Spectrum of Congenital Anomalies from Sub-Saharan Africa: Zambia-Prehistorical times I

Am J Phys Anthropol. 1999 Mar;108(3):311-9.

A prehistoric example of polydactyly from the Iron Age site of Simbusenga, Zambia.

Murphy KA[1].

Abstract

Human burials, dated AD 1100-1500, were examined from the Iron Age site of Simbusenga, located some 35 miles northwest of Victoria Falls in Zambia. Pedal polydactyly was discovered in the fragmentary remains of a young adult of indeterminate sex aged 14-25. The preaxial form of polydactyly is indicated with bilateral involvement of the first metatarsals. There is incomplete hypoplastic duplication of both first metatarsals with broad heads for the metatarsal-phalangeal joints. No digital malformations were found in the other seven individuals with feet and/or hands from the site. Several studies point to autosomal dominance for cases of isolated

Spectrum of Congenital Anomalies from Sub-Saharan Africa: Zambia-Current Profiles II

Med J Zambia. 1977 Oct-Nov;11(5):127-38.

Major congenital malformations in neonates at U.T.H. Lusaka Zambia.

Sukhani S, Patel YK, Chintu C.

Abstract

The incidence of major congenital malformations in U.T.H., Lusaka during 1976 is reported. The incidence of some common major malformations are compared with the other series. Central nervous system and Alimentry system malformations were most frequent. The relative low incidence of anencephaly and cleft lip and palate in African newborns is confirmed.

Spectrum of Congenital Anomalies from Sub-Saharan Africa: Zambia-Current Profiles III

World J Surg. 2012 Feb;36(2):241-6. doi: 10.1007/s00268-011-1158-8.

Plastic and reconstructive surgery in Zambia: epidemiology of 16 years of practice.

Jovic G[1], Corlew DS, Bowman KG.

The epidemiology of surgical conditions in developing countries is not well studied, but plastic and reconstructive surgery can play a significant role in meeting the need for surgical care. Knowledge of the conditions treated by a plastic surgeon in a low-income country would inform the development of surgical services.

METHODS:

The surgical log of the lead author from 1993 to 2008 was reviewed. The cases were performed in 33 surgical facilities in Zambia, and name, gender, age, diagnosis, procedure, and hospital were prospectively recorded. Data were analyzed for the number and distribution of cases and for patterns related to age and gender.

RESULTS:

Between 1993 and 2008, 5,740 operations were performed, and complete data were available for 5,735 (99.9%) patients. There were 5,774 surgical diagnoses. Of these, 3,885 (67.2%) were acquired conditions. These included 1,985 (34.3%) burns, 514 (9.0%) keloids, 448 (7.8%) nonburn traumas, 410 (7.1%) deep tissue infections, and 343 (5.9%) tumors. The 1,889 (32.7%) congenital conditions included 1,322 (22.9%) craniofacial defects and 354 (6.1%) limb defects. Children accounted for 78.2% of burns. Trauma cases were predominantly male (273, 60.9%). Congenital conditions were repaired after 5 years of age in 355 (18.8%) cases.

CONCLUSION:

Based on a 16-year case log from one developing country, more than half of conditions related to plastic surgery comprised injuries and congenital anomalies. Age- and gender-related patterns were evident. These findings may inform the provision of resources for injury prevention, surgical training, and delivery of surgical services

Spectrum of Congenital Anomalies Reported from Selected Sub-Saharan Countries

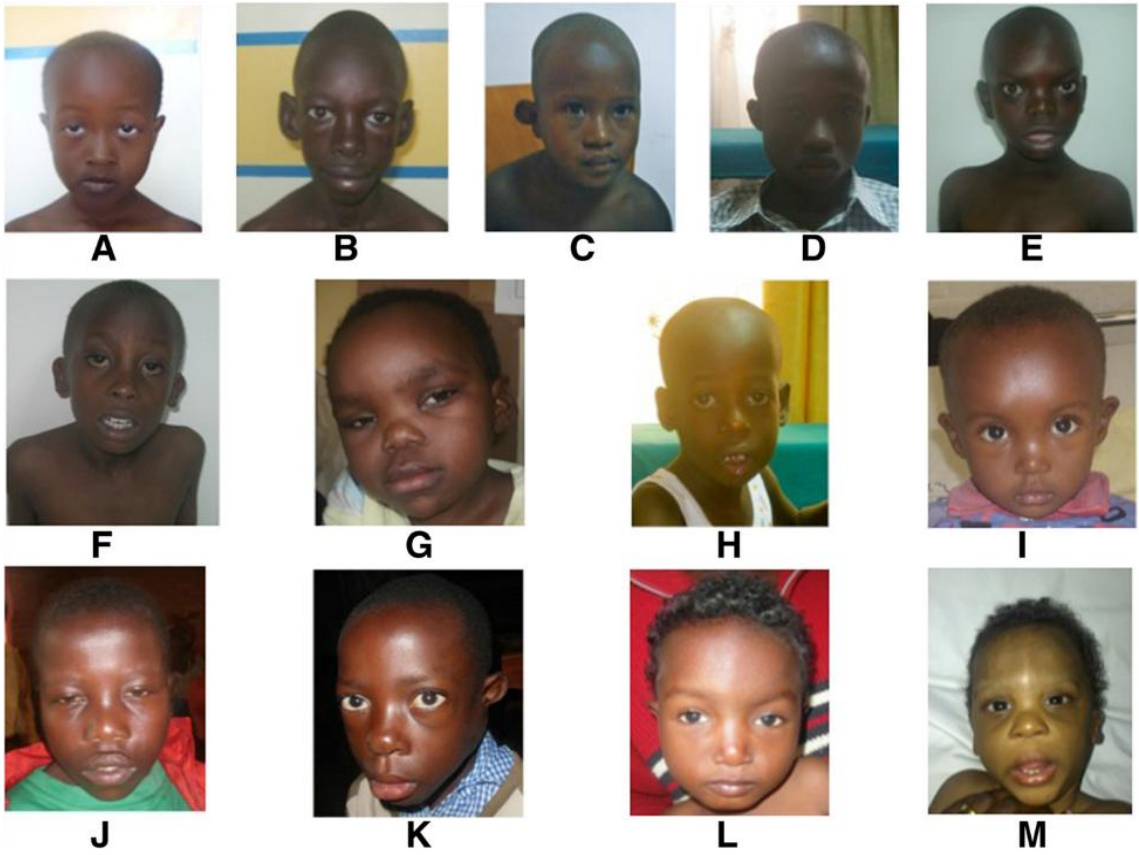
Country	Study Pop	Top 5 Congenital Defects				
Congo DRC (Kalisya LM et al.2015)	1301 (Surgical)	Talipes (35%)	Cleft lip (24%)	Inguinal Hernia (15%)	Hydrocephalus (5%)	U/Hernia (4%)
Ethiopia (Mekonen HK, et al. 2015)	1516 (Live births)	CNS (1.45%)	MSS (0.26%)	GIT (0.197%)	Others (0.13%)	GUS (0.07%)

Extended Spectrum of Congenital Anomalies from Sub-Saharan Africa: Rwanda

(Uwineza A., **Mutesa L.**, et al. BMC Medical Genetics 2014, 15:79)

Patient	Genetic Defect/Syndrome
A	trisomy 18p
B	william- Beuren syndrome
C	del 6q16.1q21
D	del 22q11.2
E	dup 1p35.3 p31.3
F	del 8p23.1
G	del 7q34q36.2
H	del 2q33.1q33.3
I	dup 7q11.23
J	dup 8q24.3/del 16p13.3
K	del 22q11.21
L	del 10p15.3p14
M	del 17q21.3q21

Array-Comparative Genomic Hybridization **(CGH)** analysis in Rwandan patients presenting development delay/intellectual disability with multiple congenital anomalies-13 of 50 Children evaluated



Challenges and Key Issues

Key Issues and Challenges

Clinical

- Awareness
- Low clinical index of suspicion
- Huge patient work load-over crowded with infectious diseases

(Difficult differential Diagnosis)

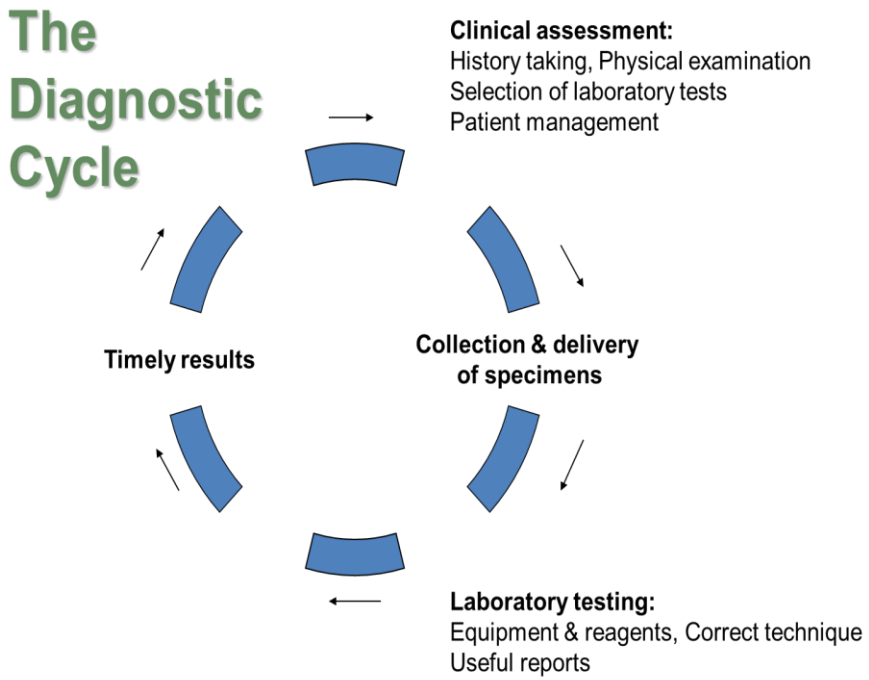
- Late Presentation
- Human resource (HR)-None/inadequate expertise

Diagnostic Support services

- None availability of equipment and diagnostic tests
- Erratic lab reagents and other diagnostics support (Immaging studies,etc)
- HR issues-lack or inadequate expertise

Cross-cutting issues

- Poverty
- Weak Health Systems



What is the Plight of a Patient with Rare Disease in Africa (Sub-Saharan Africa)?

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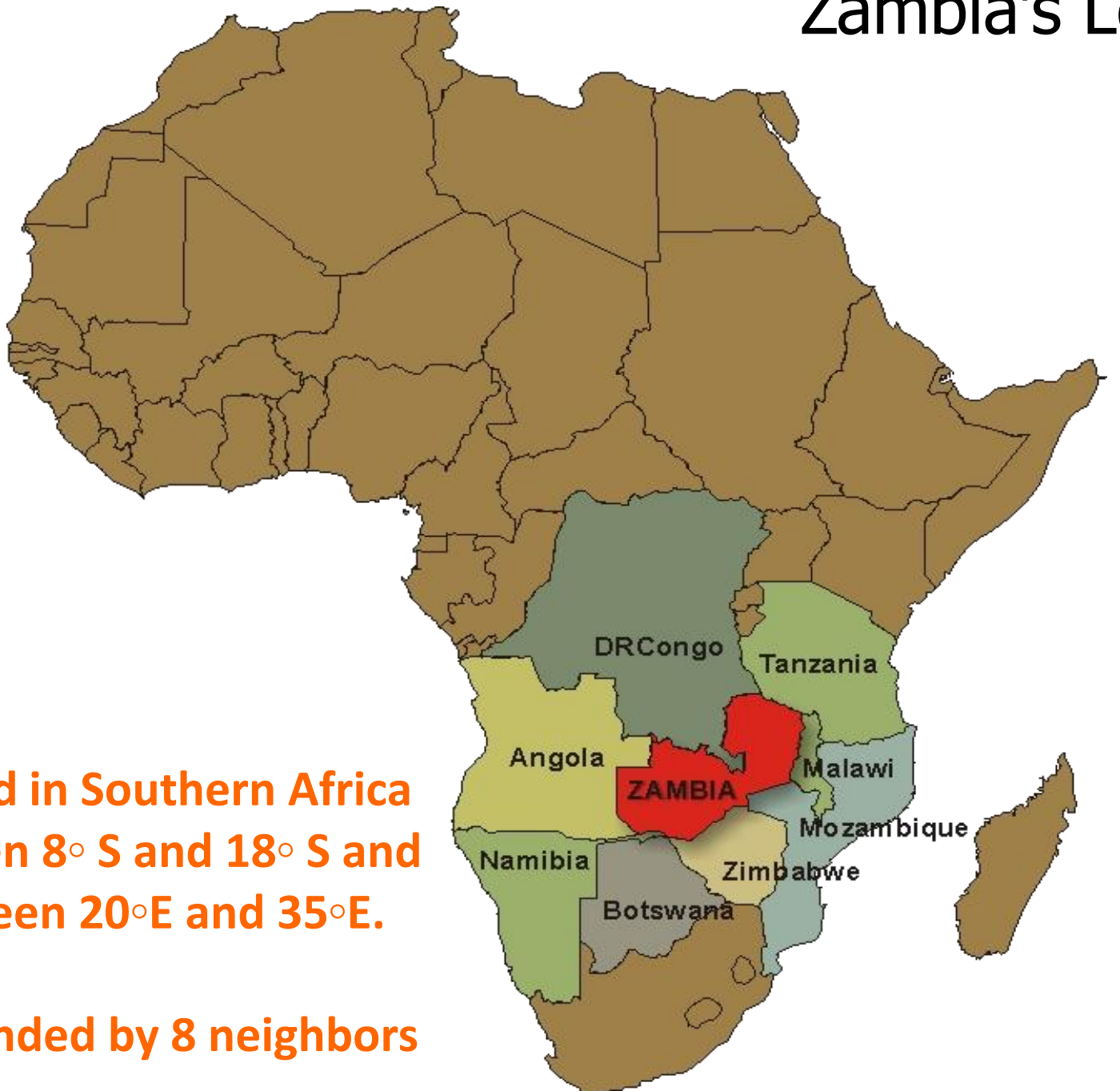
- University of Zambia School of medicine
- The School of Medicine and University Teaching Hospital Malaria Research Unit (SMUTH-MRU) team, Lusaka, Zambia

ZMA and ZPA Co-Host the 5th ASID Biannual Congress

Zambia's Location



Located in Southern Africa between 8◦ S and 18◦ S and between 20◦E and 35◦E.

Surrounded by 8 neighbors

The ICON ‘Victoria Falls’

The Victoria Falls Zambia

Unique Selling Points

The Victoria Falls Zambia

Unique Selling Points

The African Society for Primary Immune Deficiencies (ASID)

2017 5th ASID Biannual Congress

ASID Goal/Vision

ASID as a Professional Society primarily focuses on :

a) sharing experiences and raising public awareness with regard to the basic and clinical science of PIDs in Africa.

b) commitment to the promotion of evidence-based management and care of individuals suffering from PID.

c) seeking, through better and more effective interventions, to uplift the quality of life of PID patient on the African continent.

END

Thank You