Birth defects in a rural province in Papua New Guinea

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ABSTRACT

Background Globally, birth defects are the fourth most common cause of neonatal mortality. They cause substantial morbidity, and often long-term disability. Despite this, the impact of birth defects on public health has received little attention in low- and middle-income countries.

Aims To report the types, incidence and geographic distribution of birth defects in the East New Britain Province of Papua New Guinea.

Methods Data were collected over 3 years on newborns with birth defects seen at Rabaul Hospital, born anywhere in the province. Each affected newborn was examined, the anomaly diagnosed and classified. The exact home location was recorded to understand geospatial distribution. To calculate incidence, data were collected on all newborns with a congenital anomaly in a cohort of 2000 consecutive live births at Rabaul Hospital in 2019.

Results Over 3 years, 2018–2020, 137 newborns with birth defects were identified, born in any part of the province. Congenital heart defects, hydrocephalus, microcephaly, craniofacial anomalies, imperforate anus, trachea-oesophageal fistula and diaphragmatic hernia were the most common anomalies. Eight cases of Down syndrome and other chromosomal anomalies were identified. The incidence in 2019 was 14 per 1000 live births. Geographic mapping showed the highest number of cases in the region on the Gazelle Peninsula, the area around the active volcanos.

Conclusions This study provides insights into the incidence and types of birth defects in a rural island province and showed it was possible to map geospatial distribution to further explore epidemiology.

WHAT IS ALREADY KNOWN ON THIS TOPIC

⇒ Epidemiology of birth defects is not well described in low and middle-income (LMIC) countries.

WHAT THIS STUDY ADDS

⇒ The common types of birth defects and their incidence in a rural province.

HOW THIS STUDY MIGHT AFFECT RESEARCH, PRACTICE OR POLICY

⇒ The importance of birth defects in neonatal morbidity in LMIC, the need for programs to address preventable and correctable lesions, and to support children with chronic illness and disability.

⇒ Geospatial mapping can help understand disease distribution.

INTRODUCTION

Birth defects, structural and functional physical changes that are present at birth, are the fourth most common cause of neonatal mortality in low and middle-income countries (LMIC), after prematurity and low birth weight, perinatal asphyxia and infections.[1] Papua New Guinea (PNG) like most LMIC lacks adequate data regarding birth defects. Birth defects have received little attention despite their impact on the health and development of children worldwide. Most LMIC lack registries and data regarding types and prevalence of birth defects, which could prove useful for raising awareness and guiding policies for national birth defects programmes. Far from being conditions for which nothing can be done, a significant proportion of birth defects can be prevented and treated, and consequent disability reduced. For these reasons, we sought to understand the epidemiology of birth defects in PNG.

Since 2010, in PNG, numbers of selected birth defects are recorded in the Paediatric Hospital Reporting programme and reported in the Annual Morbidity and Mortality Reports produced by the Paediatric Society of PNG.[2] For example, in 2019, 275 newborns were reported with congenital malformations, of which 89 had multiple defects with an overall case fatality rate of 29.5%. However, these are a minority of all birth defects that occur in PNG, and these data do not enable an understanding of incidence or other epidemiological aspects.

One published hospital-based study[3] and two unpublished studies[4 5] have reported birth defects in PNG. The incidences reported (per 1000 live births) were 12 in 1985,[3] 8 in 1987–1996[5] and 28 in 2018.[4] There have been no studies looking at population-based data or geographical distribution of birth defects in PNG.

METHODS

We conducted a 3-year study of newborns delivered at, referred to, or brought by parents or caregivers to Rabaul Hospital between 1 January 2018 and 31 December 2020, to describe and classify the types of birth defects and to map the geospatial distribution of birth defects in the province. Because the population-numerator is uncertain (ie, many births occur in villages and are unrecorded), in a consecutive cohort of the first 2000 live births at Rabaul Hospital in 2019, we calculated the incidence of birth defects.

Included in this study were all newborns with a birth defect with gestational age of 28 weeks or more, and weight of 1000 g or more.

Thee following data collection and case ascertainment tools were used to diagnose and classify birth defects:

1. WHO Birth Defects Surveillance: quick reference handbook of selected birth defects and infections.[6]
2. ICD-10 code Q80-89 classification of congenital malformation syndromes with anomalies occurring in multiple systems.[7]
3. Comprehensive New Born Screening: handbook for screening visible birth defects at all delivery points.[8]
4. Birth Defects Surveillance: a manual for programme managers second edition.[1]

We designed forms that were completed as cases were identified and we gathered data from the special care nursery admission register, and the labour ward birth register. An electronic version of a proforma was designed using Adobe LiveCycle Designer ES V.8.2 to record patient information.

We describe the types and geographic distribution of cases of birth defects in East New Britain Province. The cases were mapped using the following steps. The primary place of residence of the mothers during their pregnancy was determined and classified according to the census division of wards. Coordinates of wards (local level government) were determined using geodata reference tables for East New Britain Province. An Excel spreadsheet of the co-ordinates was created and exported to Quantum Geographic Information System (QGIS, https://qgis.org/en/site/), which is publicly accessible downloadable software. The QGIS software was used to plot all cases on a map of East New Britain. Data on the distribution of volcanic activity were gathered from the Global Vulcanism Project at the Smithsonian Institute.[9]

To explore apparent geospatial patterns, we used the number of birth defects identified from each district, in relation to the estimated district populations from most recent PNG census. These 2011 census estimates had been extrapolated to estimates of expected population in 2019 by the PNG National Health Statistics Office, as there were no more recent other population data.[10]

The study was approved by the Rabaul Provincial Hospital Administration and Clinical Governance Committee.

RESULTS

In the 3 years, 2018 to 2020, 137 newborns with birth defects were seen at Rabaul Hospital, all born in East New Britain Province (table 1, and online suplemental file). Congenital heart defects, hydrocephalus, microcephaly, craniofacial anomalies including cleft lip and palate, imperforate anus and other obstructive lesions of the gastrointestinal tract, trachea-oesophageal fistula and diaphragmatic hernia were the most common anomalies. Eight cases of Down syndrome, plus other chromosomal anomalies, were identified.

Among 2000 consecutive live births at Rabaul Hospital in 2019, 28 infants had birth defects, giving an incidence of 14 per 1000 live births.

The home location of each of the 137 affected newborns is mapped in figure 1. Cases appeared to be more concentrated near interdistrict borders and mountain ranges along the Gazelle Peninsula (figure 2), where the Rabaul volcanos are active. The distribution of volcanic activity in the Gazelle Peninsula is included in figure 2.[9]

Table 2 explores the incidence per estimated district population. Based on these data, there was no evidence of disproportionate incidence of cases of birth defects relative to district population estimates.

Table 1 Birth defects seen at Rabaul Hospital 2018–2020 born at any health facility or born in villages in East New Britain Province

Type of birth defect	Number (%)
Congenital heart disease	**37 (27.0)**
Central nervous system anomalies	**19 (13.9)**
Hydrocephalus	5
Microcephaly	5
Anencephaly	3
Cervical meningocoele	2
Craniofacial anomalies	**19 (13.9)**
Cleft lip and palate	8
Isolated cleft soft palate	2
Isolated cleft palate	1
Craniofacial anomalies—cleft palate, microtia, facial hypoplasia	1
Choanal atresia	1
Laryngomalacia	3
Microphthalmia	1
Pierre Robin sequence	2
Chromosomal and identifiable genetic syndromes	**26 (19.0)**
Down syndrome	8
Patau syndrome	4
Treacher-Collins syndrome	1
Undiagnosed chromosomal syndrome	13
Gastrointestinal system and abdominal anomalies	**14 (10.2)**
Imperforate anus	8
Duodenal atresia	1
Caecal stenosis	1
Gastroschisis	1
Omphalocele	1
Umbilical hernia	2
Respiratory and thoracic anomalies	**12 (8.7)**
Tracheo-oesophageal fistula	5
Diaphragmatic hernia	4
Laryngomalacia	3
Musculoskeletal and limb anomalies	**6 (4.4)**
Skeletal dysplasia	1
Syndactyly	1
Talipes	3
Renal and genitourinary anomalies	**4 (2.9)**
Hydrocoele/hypospadias/micropenis/phimosis	4
Congenital malformation syndromes affecting multiple systems	**7 (5.1)** *
Total	**137**

*Based on ICD-10 code Q80-89 classification of congenital malformation syndromes with anomalies occurring in multiple systems.[7] These seven cases are also listed according to individual systems affected. Includes one case of congenital rubella syndrome.

DISCUSSION

This study provides insights into birth defects in the East New Britain Province of PNG. The incidence was 14 per 1000 live births. The most common types of birth defect identified were congenital heart defects, hydrocephalus and microcephaly, craniofacial anomalies and obstructive lesions of the gastrointestinal tract. This incidence is similar to that reported in other studies in PNG in the last 30 years.[3–5] Although in high-income countries hospital-based data overestimates the incidence of congenital anomalies at a population level, in rural PNG, as in many low-income settings, there is no antenatal diagnosis of birth defects, theerefore referral bias is absent. There may be a

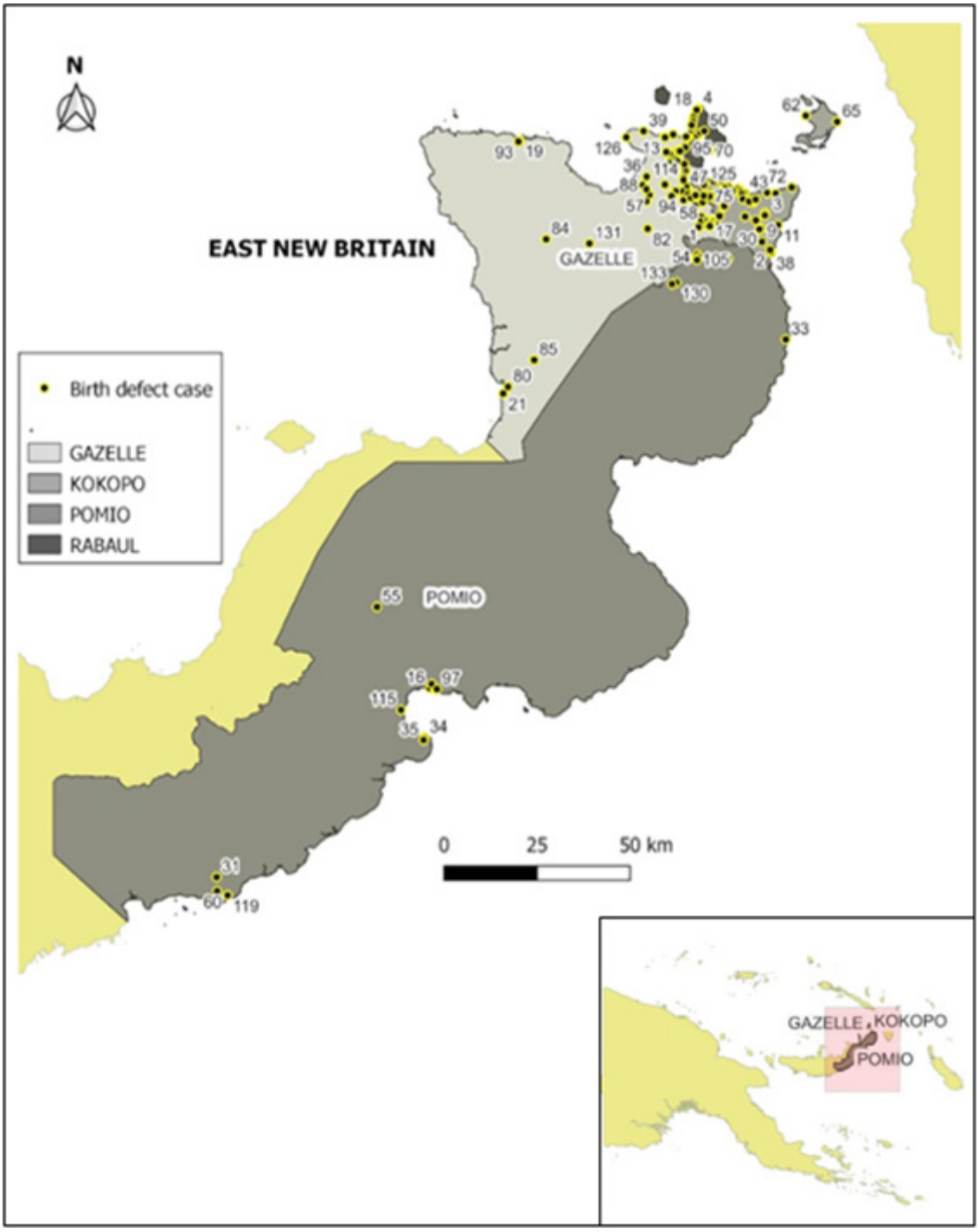

Figure 1 Geospatial mapping of birth defects identified over a 3-year period in the four districts of East New Britain Province. Numbers are birth defect case numbers.

small bias from antenatal referral of mothers for maternal high risk, which may in turn be associated with a higher risk of birth defect in a newborn, but this effect is very small in such settings. The hospital acts as a community hospital, so the 2000 consecutive births at the hospital reflect similar births in the catchment community.

It is more likely that we *underestimated* the *number* of birth defects throughout the province in the 3-year period we evaluated. In East New Britain, 40% of mothers deliver outside the main health facilities in the province, which is why we could not calculate an incidence for the entire 3-year cohort.[11] We almost certainly underestimated infants born with birth defects in more remote parts of the province, as some affected newborns will not be brought to hospital or may die before they can be brought to hospital.

Finding of cases apparently concentrated along the Gazelle Peninsula was not expected. This may reflect access bias, that is the population of mothers bringing their newborns to Rabaul Hospital will be influenced by geographical access, as is referral of newborns recognised as having birth defects in remote health facilities. It was difficult to investigate this apparent clustering based on incidence per district population, as the Gazelle

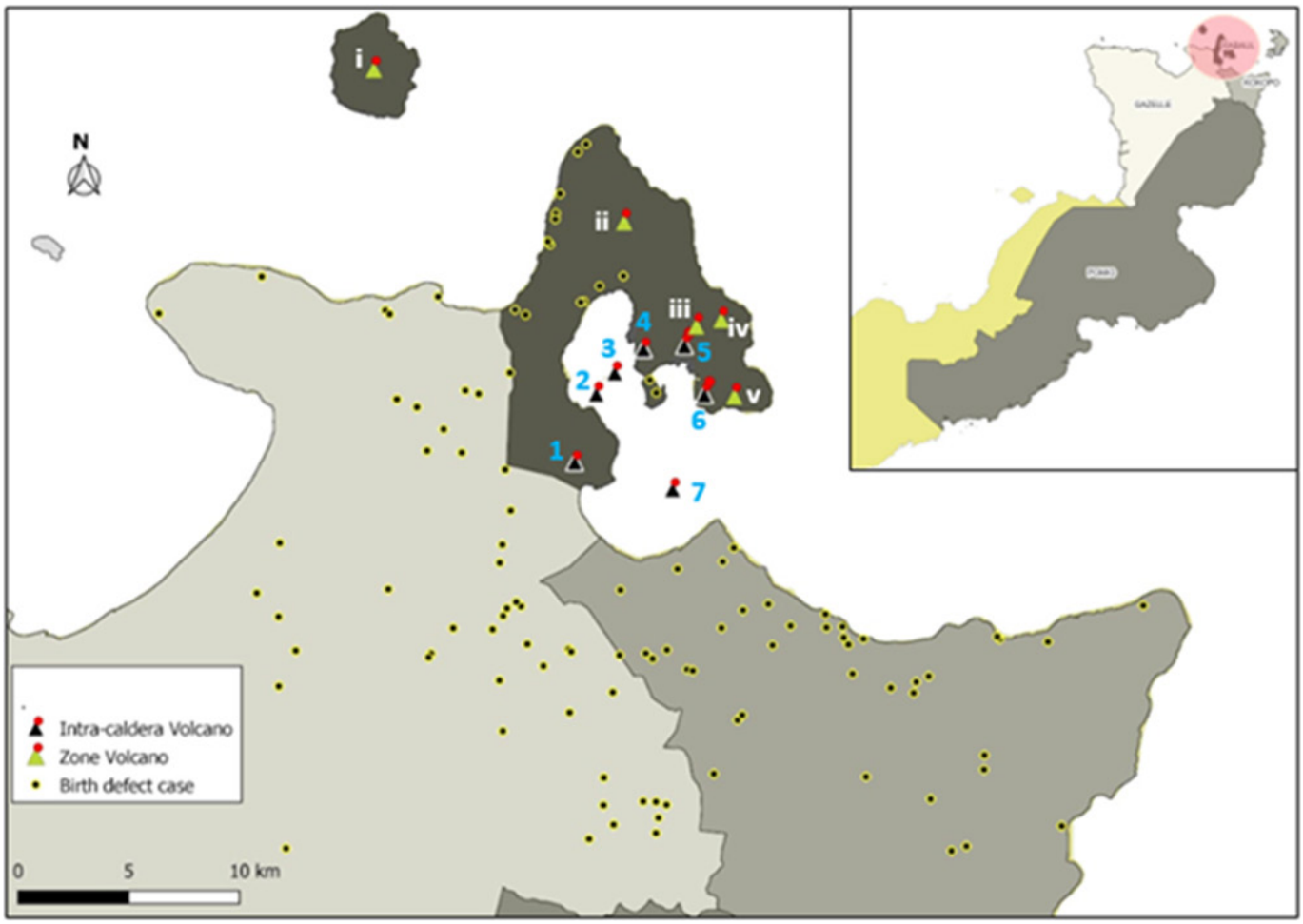

Figure 2 Geospatial mapping of birth defects identified over a 3-year period with focus on the region of the Gazelle Peninsula. Numbers are birth defect case numbers. The volcanic regions of the northern part of the Gazelle Peninsula lie within the Pacific Ring of Fire.[9] These include the Rabaul Caldera and the Watom-to-Turagunan Zone with either landform or submarine volcanoes that are active, dormant or extinct after previous activity. The Rabaul Caldera has seven intra-caldera volcanoes—1. Vulcan, 2. Un-named, 3. Dawapia Rocks, 4. Sulphur Creek, 5. Rabalanakaia, 6. Tavurvur and 7. Karavia Bay caldera. The Watom-to-Turagunan Zone has five zone volcanoes—i. Watom, ii. Tovanumbatir, iii. Palangiangia, iv. Kabiu and v. Turagunan.

Peninsula overlaps more than one district (the north-eastern tip of Gazelle district and all of Kokopo and Rabaul districts), and population estimates of the Gazelle Peninsula per se do not exist. The only data we had on population, represented in table 2, did not support a clustering.

Table 2 Distribution of birth defects by district over 3 years (2018–2020)

Districts	Number of cases observed (% of all cases)	District population (% of district population in the province) in 2019 *	Population density (population per km^2 [10]	Estimated incidence of birth defects per 1000 population
Gazelle	58 (42)	149 538 (41)	35	0.39
Kokopo	44 (32)	97 184 (27)	220	0.45
Pomio	19 (14)	74 896 (20)	6.5	0.25
Rabaul	16 (12)	45 005 (12)	410	0.36
Total	**137 (100)**	**366 623 (100)**		**0.37**

*Population estimates for 2019 extrapolated by the National Statistics Office from 2011 census data.

However, causes of potential clustering in this region, particularly environmental factors, warrant consideration. The Gazelle peninsula surrounds the active Rabaul volcanos, and cadmium is a heavy metal that is found in high concentrations in volcanic ash emissions. On the Gazelle Peninsula, significant levels of cadmium have been detected in the local food chain.[12] As an environmental toxin cadmium accumulates in marine ecosystems,[13] food chains and within living organisms. Cadmium exposure in humans has been linked to a higher risk of preterm delivery,[14] congenital heart disease,[15] cleft palate and other teratogenic effects as well as chronic adult diseases.[16] In 2013, an agricultural study in the East New Britain Province identified high levels of cadmium in vegetables, nuts, marine organisms and tobacco. There was evidence of significant levels of cadmium in the food chain around the volcano, and that study proposed it may be associated with adult diabetes and hypertension seen in East New Britain.[12]

Our study cannot shed more light on whether the apparent distribution of birth defects resulted from differences in population density, access to the provincial hospital, referral bias, environmental exposure related to the volcano or other unknown factors. However, there is a need for further intersector research

into cadmium and other volcanic toxins to inform public health policies.

In PNG and other LMIC, tools and guidance are needed for identification of birth defects to aid diagnosis by primary healthcare workers, reporting and research. Healthcare workers need referral guidelines for birth defects, as many are treatable, or managed in a way that reduces disability. While national population-based surveillance may not be feasible at this time, sentinel hospital-based surveillance via the Paediatric Hospital Reporting system is possible and increased awareness and reporting of birth defects through this programme is needed.[2]

Although we cannot quantify this from our data, some birth defects are preventable, and there is a need for feasible approaches in low resource settings: prevention of sexually transmitted diseases, vaccination against rubella and fortification of staple foods with folic acid, iodine and other micronutrients as well as investigation and legislation controlling management of toxic chemicals. These approaches should be coupled with strengthened prenatal and perinatal services, including promotion of family planning, expanding rubella immunisation before pregnancy and implementing folic acid fortification of commonly consumed foods. We could not test for rubella, zika virus or other congenital infections, but note there were five cases of microcephaly, and one case that fulfilled clinical criteria for congenital rubella syndrome.

There should be training of healthcare professionals in best practices in the care and prevention of birth defects and in the practical application of medical genetics, at both the undergraduate and postgraduate levels. Principles of genetic counselling can also be taught to nurses and doctors in training.

Education of the community is also needed on the steps that can be taken to promote healthy outcomes of pregnancy, including the avoidance of potential teratogens during gestation, particularly alcohol, and on the importance of knowing how and when to access prenatal care.

The encouragement of parent and patient organisations to help each other and advocate for better services for children with birth defects is important. Parent groups can draw attention to the need for improved clinical, educational and social services for children with congenital and acquired disability to assert their right to be treated with dignity and without discrimination.

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Competing interests None declared.

Patient consent for publication Not applicable.

Ethics approval This study involved human participants and was approved by Rabaul Provincial Hospital Administration and Clinical Governance Committee. Participants gave informed consent to participate in the study before taking part.

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