CASE REPORT

Myxoedema in a patient with achondroplasia in rural area of Guatemala

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SUMMARY

A 43-year-old indigenous Guatemalan woman with achondroplasia presented to our clinic with chronic fatigue and generalised oedema. She had limited contact with the formal healthcare system. However, 1 year prior, she had sought medical evaluation from a private physician. Her symptoms had been attributed to a combination of heart failure and physical disability due to the musculoskeletal complications of her achondroplasia. She was lost to follow-up due to inability to pay for further testing or treatment. On initial laboratory evaluation in our clinic, she was found to have a thyrotropin level greater than assay. With initiation of oral levothyroxine supplementation, her dyspnoea and oedema completely resolved. The case illustrates how indigenous patients in rural Guatemala experience many barriers to accessing high-quality medical care. As a result, presentations of common illnesses are often very advanced and definitive diagnoses and treatments are frequently delayed.

CASE PRESENTATION

A 43-year-old Guatemalan woman with achondroplasia, of indigenous Maya ethnicity and a monolingual speaker of K'iche' Maya was brought by her mother for evaluation in our outpatient clinic located in a rural agricultural area in the Piedmont region of Guatemala. The patient endorsed more than 1 year of gradually progressive symptoms, including dyspnoea with exertion, orthopnoea, fatigue, anorexia, cold intolerance and generalised oedema.

Previously, the patient had had limited contact with the formal healthcare system. On interview, with the assistance of our K'iche'-speaking nursing staff, her mother explained that she had experienced a normal, full-term vaginal home birth but that within the first year of life she noted that the patient had short arms and legs and appeared to develop slowly. She took her to the local health post where she was told that her child was 'special' and that 'nothing could be done for her'. Subsequently, when the patient reached school age, the patient and mother reported that she experienced bullying and stigma and was discouraged by the local public school teachers from attending school. Nevertheless, at home, she participated fully in family life and enjoyed cooking, cleaning and other domestic chores until the onset of her current symptoms about 1 year ago.

When her symptoms began 1 year prior, she and her family were initially reluctant to present to care because of prior stigmatising experiences related to

the patient's achondroplasia. However, as symptoms progressed, they decided to use some of their savings to seek a consultation with a private generalist physician. That physician had recommended an echocardiogram (ECG), which they had paid for out of pocket at considerable expenses. At a follow-up visit, they reported to us that they had been told that 'something was wrong with her heart' but that they had difficulty understanding what they were told. They were largely monolingual K'iche' Maya speakers, and the clinic staff had only spoken Spanish. Regardless, they had been told that additional testing and medications would be necessary, and so they declined further follow-up as they had exhausted their available funds on the echocardiogram. Her symptoms had continued to progress throughout the year.

On examination, the patient appeared fatigued. She had mild resting tachypnoea which worsened with any exertion. Her heart rate was 60 and regular, arterial blood pressure was 90/50 mm Hg, weight was 27.7 kg, height was 95 cm and body mass index was 30.7. She had generalised pallor and dry skin. On auscultation, she had a 2/6 holosystolic murmur at the left lower sternal border that augmented with inspiration and bibasilar inspiratory pulmonary crackles. Her abdomen was soft with a reducible umbilical hernia. She exhibited generalised pitting oedema and had diminished deep tendon reflexes. She exhibited extreme short stature, frontal bossing of the forehead, rhizomelic shortening of the arms and legs and exaggerated lumbar lordosis.

The patient brought a copy of her echocardiogram from the prior year which we reviewed. Major findings included mild left ventricular concentric hypertrophy and diastolic dysfunction, tricuspid regurgitation with a pulmonary artery systolic pressure of 40 mm Hg plus right atrial pressure and small pleural and pericardial effusions.

Laboratory testing was performed in our clinic. Significant results included serum levels of thyrotropin >75 µUI/mL (0.4–4.0 µUI/mL), haemoglobin 8.8 g/dL (11–18 g/dL), haematocrit 29% (35–60%), creatinine 0.8 mg/dL (normal range 0.60–1.10 mg/dL), potassium 4.7 mmol/L (3.5–5.3 mmol/L), sodium 131 mmol/L (135–148 mmol/L) and normal urinalysis.

The patient was initiated on oral levothyroxine supplementation at 25 µg/day, with subsequent titrations at 4–6-week intervals based on repeat serum thyrotropin measurements. With treatment, the patient's symptoms of dyspnoea, fatigue and oedema resolved completely.

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GLOBAL HEALTH PROBLEM LIST

1. In Guatemala, there is pervasive social stigma and limited access to services for those with disabilities.
2. Individuals who are indigenous Maya or live in rural areas experience many significant barriers to accessing high-quality care, including the high cost of care and language barriers for those who speak a Mayan language.
3. Epidemiology of common chronic illnesses, including acquired hypothyroidism, is limited in Guatemala.

GLOBAL HEALTH PROBLEM ANALYSIS

This case describes an indigenous Guatemalan woman who manifested an advanced presentation of a common illness (acquired hypothyroidism). Severe delays in diagnosis and treatment of this illness were occasioned by her inability to pay for diagnostic testing and treatment; by her difficulty with comprehending medical information due to language barriers and by her prior experiences of stigmatisation related to a coincident uncommon disease (achondroplasia).

Social stigma related to disability: Our patient and her family were reluctant to present to medical care because she had experienced severe stigma in her childhood related to her achondroplasia. Indeed, she had had no formal education because she had been excluded from public schooling due to her condition. Furthermore, she had had no significant contact with the health-care system from childhood until she became ill with acquired hypothyroidism.

Although Guatemala is a signatory to the *Convention of the Rights of Persons with Disabilities*,^{1 2} there remain few social services available for individuals with disability. Individuals with disability are often de facto excluded from the public educational system, as was our patient. Furthermore, given limited physician exposure to and training in disability medicine, individuals with disability are often told that ‘nothing can be done for them’—as was our patient early in her life—which leads to disillusionment with and disengagement from the medical system.³ Another important barrier for the patient and her family, limiting their access to medical care and other social services, was difficulty with accessing public transportation, which makes no special accommodations for individuals with disability.

Reversing these disparities requires a multisectorial approach. However, one important step forward will be the collection of epidemiological data on the burden of disability in Guatemala. Currently, such data are almost completely lacking. For example, although achondroplasia is the most common cause of dwarfism worldwide,⁴ we could find no clinical reports or epidemiological studies for the condition in Guatemala—even though, somewhat ironically, such reports do exist in the Guatemalan archaeological literature.⁵

Importantly, along these lines, a new National Disability Survey in Guatemala is currently underway, and we eagerly anticipate these results. As a preliminary effort prior to the release of the full survey, Guatemala’s first directory of public sector and civil society providing services to individuals with disabilities has just been released.⁶ One major feature of this directory is that it highlights the important role that self-affiliating patient and family associations are playing in the current advocacy climate in Guatemala. For example, the Gente Pequeña de Guatemala (Little People of Guatemala) Association is a major force in advocating for the rights of Guatemalan individuals with achondroplasia.⁷ Another important recent development is a report issued during a recent visit to Guatemala by the United Nations Committee on the Rights of Persons with Disabilities, which made several strong recommendations for revised national

legislation and policy framework.⁸ Civil society and disability advocacy organisations, such as those mentioned above, can assist with the implementation of these recommendations by advocating with elected government officials to implement the required legislative changes.

Finally, our case highlights the need for broader changes in focus at the primary healthcare level in Guatemala—and in other low-income and middle-income countries as well—to include an emphasis on early child development. The moment in our case report where the patient’s mother was told that ‘nothing could be done for her’ because she was ‘special’ highlights a lack of capacity to think broadly about fostering child development, rather than just focusing narrowly on physical growth as the only metric of interest. Although training in early child development is virtually non-existent in Guatemala at this time, very recent international policy emphases on early child development⁹—as well as new training materials for frontline health workers produced by the WHO¹⁰—should lead to new programmatic emphases in public and private sector efforts to integrate training in child development and developmental services into existing primary care programmes.¹¹

Barriers to care for indigenous persons: Guatemala has one of the largest indigenous populations in Latin America, nearly 50% of the total population.¹² Around the world, indigenous people suffer from significant economic and healthcare disparities^{13 14} and Guatemala is no exception. Several scholars have documented the ways in which discrimination against Guatemala’s indigenous population leads to significant disparities in economic prospects, health outcomes and access to basic infrastructure.^{12 15–17} These disparities are also well documented in representative national surveys on maternal-child health outcomes and living conditions.^{18 19} One important determinant of these disparities is the fact that many indigenous persons primarily speak one of Mayan languages and have limited fluency in the dominant Spanish.²⁰ Although Guatemala has passed legislation guaranteeing access to healthcare and other social services in indigenous languages, in practice there are few resources available to assist with implementing or enforcing the provisions of these laws.²¹ In practice, this means that many indigenous Maya individuals are reluctant to present for medical care and, when they do, may not be able to fully grasp the recommended diagnostic or treatment plan.

Another significant barrier for indigenous persons, who suffer from rates of poverty nearly double than those of their non-indigenous counterparts in Guatemala,²² is the high cost of medical care. In Guatemala, healthcare for chronic diseases has become increasingly privatised since the 1990s. This is because the public health system is underfunded,¹² in part due to the influence of international priorities in healthcare financing. In particular, large-scale donors in global health have primarily funded maternal-child health and infectious disease programmes, with <2% of all international healthcare funding directed towards chronic illnesses.²³ As national governments—including the government of Guatemala—rely on these donor sources to build new healthcare initiatives, there is a palpable lack of capacity for managing chronic illnesses. As a result, most chronic disease care must be paid for out of pocket.²⁴ As such, the story of our patient is in many ways typical. She exhausted all her savings on her initial medical consultation and unrevealing echocardiogram and therefore was unable to pay for the additional recommended analytical blood testing which would have revealed the diagnosis.

Although these structural problems of social inequality in Guatemala do not have simple solutions, a few practical considerations for healthcare organisations and providers working in

rural Guatemala may help to mitigate them. First, organisations and providers must begin to demonstrate their commitments to complying with Guatemala's legislation on indigenous languages by ensuring that their patients have access to interpreter services. Unlike many high-income countries, interpretation is still an informal, unprofessionalised field in Guatemala without formal training programmes or certification standards.²⁵ Nevertheless, employing and training indigenous healthcare providers—such as nurses—to fill this role is a promising strategy and, indeed, is the strategy we employ in our clinics, such as the one where the patient discussed here presented for care. Recently, we have described the training programme we have developed to teach these essential interpretation and care navigation skills to our healthcare providers.²⁶

Second, healthcare providers must be exceedingly conscious of the out-of-pocket costs their rural Guatemalan patients incur when following diagnostic and treatment plans. In the case of our patient, for example, the differential diagnosis for her initial presentation was broad, and included heart failure, malnutrition, chronic kidney disease or the nephrotic syndrome and hypothyroidism.^{27–28} Proceeding in a cost-conscious, sequential fashion through indicated diagnostic tests may have led to a timely diagnosis. However, because the initial test chosen was a quite expensive one (out-of-pocket cost for echocardiogram ~US\$100), the patient exhausted her funds and was unable to continue in care (cost of a serum thyrotropin assay, ~US\$12).

At a larger structural level, developing mechanisms for financing chronic disease care are urgently needed. At a public policy level, the recently formed WHO Global Coordination Mechanism has brought together high-level officials from numerous ministries of health to discuss possible financing mechanisms. Major recommendations for individual countries include raising the tax base for chronic disease funding (eg, through taxing products like tobacco) and directing international development assistance funding towards chronic disease programming.²⁹ This latter strategy will require consistent advocacy by governments and civil society towards major international donor agencies in order to redirect their funding priorities towards chronic diseases.³⁰

A final financing mechanism recommended by the WHO commission is continued mobilisation of alternative community-based resources.²⁹ As an example of this approach, mutual health organisations or mutual health insurances have recently become a popular financing solution in some low-income and middle-income countries, especially in Africa.^{31–32} These mechanisms have not been well explored in Latin America, however, and so further research to investigate their feasibility in Guatemala and other Latin American countries will be important.

Epidemiology of chronic illnesses in Guatemala: Guatemala is rapidly undergoing a transition towards a higher burden of non-communicable diseases. Nevertheless, as this case illustrates, the healthcare system has limited capacity for diagnosing or treating these illnesses. Especially for the indigenous population, epidemiological data on non-communicable diseases to help guide policy and practice is almost completely absent.

In the case of thyroid disorders, Guatemala has a long tradition from the last century of participating in formative epidemiological and public health investigations. For example, as early as the 1950s, researchers in Guatemala were concerned with the epidemiology of endemic goitre, which at that time affected more than 50% of the rural adult population.³³ Indeed, Guatemala was one of the first countries in Latin America—and the first in Central America—to pass legislation requiring

iodisation of table salt.³⁴ Interest in public health interventions has waned however and have been difficult to sustain due to lack of funding. For example, a pilot newborn screening programme in one of Guatemala's largest public hospitals demonstrated that the incidence of congenital hypothyroidism was up to three times higher than that reported in many other countries, but the programme was abandoned due to funding cuts.³⁵ In the case of acquired hypothyroidism—such as that of our patient, which presents with adult-onset symptoms in the absence of goitre—no published reports or clinical guidelines are available. In the absence of such data, brief reports, such as this one, can assist rural healthcare providers by describing case presentations and treatment strategies used, and by closely analysing the barriers to care which our patients routinely encounter. Ultimately, as explored in the prior two sections, improving funding for chronic diseases and care delivery for individuals with disabilities will simultaneously improve access to meaningful epidemiological data on chronic diseases. This will further help to guide policy and resource allocation decisions.

Patient's perspective

'When I had to go downtown, I had to take a moto-taxi because it was hard for me to take the bus. And when I had to go to the hospital, I had to go in car, because I had too much pain, and the movement of the bus worsens my pain. But now I feel better, it is easier to take the bus to the market because I don't have pain in my body and no fever'.

Learning points

- ▶ Stigma is a persistent social problem for individuals with disabilities in Guatemala. This limits their willingness to access medical care and may contribute to late, advanced presentations of concomitant chronic illnesses.
- ▶ Poverty and language barriers are barriers for indigenous persons, which further contribute to delayed diagnosis and treatment of non-communicable diseases.
- ▶ In a healthcare system oriented primarily towards the treatment of acute illnesses, the high out-of-pocket costs for the treatment of chronic illnesses limit access to care.
- ▶ Distance and transportation limit access of individuals with disabilities to health services.
- ▶ Myxoedema is an uncommon advanced presentation of severe hypothyroidism which must be considered in the differential diagnosis of adult-onset fatigue and generalised oedema.

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Patient consent Obtained.

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REFERENCES

- 1 UNICEF. *The state of the world's children 2013: children with disabilities*. New York, NY: UNICEF, 2013.

- 2 UN Committee on the Rights of Persons with Disabilities. *Observaciones finales del comité sobre los derechos de las personas con discapacidad sobre el informe inicial de Guatemala*. Geneva, Switzerland: United Nations, 2016.
- 3 Grech S. Living with disability in rural Guatemala: exploring connections and impacts on poverty. *International Journal of Disability, Community, & Rehabilitation* 2008;7. (cited 9 Oct 2016). http://www.ijdc.ca/VOL07_02_CAN/articles/grech.shtml
- 4 Pauli RP. Achondroplasia. In: Pagon RA, Adam MP, Ardinger HH, *et al.* eds. *GeneReviews*®. Seattle, WA: University of Washington, 1993–2016. (cited 9 Oct 2016). <https://www.ncbi.nlm.nih.gov/books/NBK1152>
- 5 Rodríguez CA, Isaza C, Pachajoa H. Achondroplasia among ancient populations of mesoamerica and South America: iconographic and archeological evidence. *Columb Med* 2012;43:212–15.
- 6 Consejo Nacional para la Atención de las Personas con Discapacidad. *Directorio de servicios para personas con discapacidad*. Guatemala City, Guatemala: Consejo Nacional para la Atención de las Personas con Discapacidad, 2016.
- 7 Pérez D. Ellos son gente pequeña. *Contrapoder*. 2015 Jul 15 (cited 9 Oct 2016). <http://contrapoder.com.gt/tag/gente-pequena-de-guatemala/>
- 8 Castaño CL. La ONU pidió a Guatemala revisar su legislación sobre los derechos de las personas con discapacidad. *Panorama* 2016. (cited 8 Nov 2016). <http://panorama.ridh.org/la-onu-pidio-a-guatemala-revisar-su-legislacion-sobre-los-derechos-de-las-personas-con-discapacidad/>
- 9 Richter LM, Daelmans B, Lombardi J, *et al.* Investing in the foundation of sustainable development: pathways to scale up for early childhood development. *Lancet* 2017;389:103–18.
- 10 World Health Organization. *Care for child development: improving the care for young children*. Geneva, Switzerland: World Health Organization, 2012.
- 11 Black MM, Walker SP, Fernald LC, *et al.* Early childhood development coming of age: science through the life course. *Lancet* 2016;389:77–90.
- 12 Chary A, Rohloff P, eds. *Privatization and the new medical pluralism: shifting healthcare landscapes in Maya Guatemala*. Lanham, MD: Lexington Books, 2015.
- 13 Gracey M, King M. Indigenous health part 1: determinants and disease patterns. *Lancet* 2009;374:65–75.
- 14 Anderson I, Robson B, Connolly M, *et al.* Indigenous and tribal peoples' health (*The Lancet-Lowitja Institute Global Collaboration*): a population study. *Lancet* 2016;388:131–57.
- 15 Cerón A, Ruano AL, Sánchez S, *et al.* Abuse and discrimination towards indigenous people in public health care facilities: experiences from rural Guatemala. *Int J Equity Health* 2016;15:77.
- 16 Poder TG, He J. The role of ethnic and rural discrimination in the relationship between income inequality and health in Guatemala. *Int J Health Serv* 2015;45:285–305.
- 17 Poder TG, He J. How can sanitary infrastructures reduce child malnutrition and health inequalities? Evidence from Guatemala. *J Dev Effect* 2011;3:543–66.
- 18 Ministerio de Salud Pública y Asistencia Social. *VI Encuesta Nacional de Salud Materno Infantil*. Guatemala City, Guatemala: Ministerio de Salud Pública y Asistencia Social, 2015.
- 19 Instituto Nacional de Estadística. *Encuesta Nacional de Condiciones de Vida 2014*. Guatemala City, Guatemala: Instituto Nacional de Estadística, 2015.
- 20 Henderson B, Rohloff P, Henderson R. Towards a development-based approach to language revitalization. *Language Documentation & Conservation* 2015;8:75–91.
- 21 Maxwell J. Revitalización de los idiomas mayas de Guatemala. In: Rohloff P, Kraemer Díaz A, Ajsivinac Sian J, eds. *Más que Desarrollo: Memorias de la Primera Conferencia Bial sobre Desarrollo y Acción Comunitaria*. Bethel, VT: Wuqu' Kawoq, 2011:177–210.
- 22 Cabrera M, Lustig L, Morán HE. Fiscal policy, inequality, and the ethnic divide in Guatemala. *World Development* 2015;76:263–79.
- 23 Dain K. Sustainable financing: the Achilles heel of the non-communicable diseases response. *Lancet Diabetes Endocrinol* 2015;3:923–5.
- 24 United States Agency for International Development. *Quién financia el sistema de salud en Guatemala?* Guatemala City, Guatemala: USAID, 2008. (cited 9 Oct 2016). http://www.proyectodialogo.org/documents/salud/politica_salud_01.pdf
- 25 Dysart-Gale D. Communication models, professionalization, and the work of medical interpreters. *Health Commun* 2005;17:91–103.
- 26 Chary A, Flood D, Austad K, *et al.* Navigating bureaucracy: accompanying indigenous Maya patients with complex healthcare needs in Guatemala. *Human Organization* 2016;75:305–14.
- 27 Ponikowski P, Voors AA, Anker SD, *et al.* 2016 ESC Guidelines for the diagnosis and treatment of acute and chronic heart failure. *Eur Heart J* 2016;37:2129–200.
- 28 Hull RP, Goldsmith DJA. Nephrotic syndrome in adults. *BMJ* 2008;336:1185–9.
- 29 WHO Global Coordination Mechanism on NCDs. *Financing national NCD responses*. Geneva, Switzerland: WHO, 2015. (cited 12 Feb 2017). <http://www.who.int/ncds/gcm/two-page-financing-NCDs.pdf>
- 30 Nugent R. *Bilateral and multilateral financing for NCDs*. Geneva, Switzerland: WHO, 2015. (cited 12 Feb 2017). <http://www.who.int/nmh/ncd-coordination-mechanism/Policybrief5.2docx.pdf>
- 31 Habib SS, Perveen S, Khuwaja HM. The role of micro health insurance in providing financial risk protection in developing countries—a systematic review. *BMC Public Health* 2016;16:281.
- 32 Awomo Ndongo JC. Mutual health organizations and health-care consumption in Cameroon. *International Journal of Health Preference Research* 2016;1:2–15.
- 33 Scrimshaw NS. INCAP studies of endemic goiter and its prevention. *Food Nutr Bull* 2010;31:111–17.
- 34 Pretell EA, Delange F, Hostalek U, *et al.* Iodine nutrition improves in Latin America. *Thyroid* 2004;14:590–9.
- 35 Chávez-Meyer H, Barreondo M, Córdón Pineda K. Tamizaje del hipotiroidismo congénito en Guatemala: incidencia durante los primeros diez años. *Pediatrica Edición Internacional* 1998;1:20–5.

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