Case report

Greenberg Skeletal Dysplasia: first reported case in the Democratic Republic of Congo

Toni Kasole Lubala¹,², Nina Lubala¹, Arthur Ndundula Munkana¹, Adonis Muganza Nyenga¹, Augustin Mulangu Mutombo¹

¹Faculté de Médecine, Université de Lubumbashi, 1825 Lubumbashi, République Démocratique du Congo, ²Centre Interdisciplinaire de Génétique au Congo, CIGEC, Lubumbashi, République Démocratique du Congo

Corresponding author: Kasole Lubala, Faculté de Médecine, Université de Lubumbashi, 1825 Lubumbashi, République Démocratique du Congo

Key words: Hydrops, skeletal dysplasia, postaxial polydactyly, Greenberg

Received: 03/11/2012 - Accepted: 09/12/2012 - Published: 11/02/2013

Abstract

We describe the first Congolese case of Greenberg Skeletal Dysplasia. Were noted at birth a congenital hydrops, a chondrodystrophy, a severe hypoplastic face as well as an ulnar (postaxial) hexadactyly on all four limbs.

Pan African Medical Journal. 2013; 14:55. doi:10.11604/pamj.2013.14.55.2170

This article is available online at: http://www.panafrican-med-journal.com/content/article/14/55/full/

© Toni Kasole Lubala et al. The Pan African Medical Journal - ISSN 1937-8688. This is an Open Access article distributed under the terms of the Creative Commons Attribution License (http://creativecommons.org/licenses/by/2.0), which permits unrestricted use, distribution, and reproduction in any medium, provided the original work is properly cited.
Introduction

Greenberg Skeletal Dysplasia or Hydrops-ectopic calcification-moth-eaten (HEM) is an autosomal recessive chondrodysplasia extremely rare characterized by fetal hydrops, short limbs and abnormal chondro-osseous calcification. Since Greenberg described the first case in 1988 [1], only about ten other cases had been reported in the literature [2]. In this article, we describe the first Congolese case of Greenberg skeletal dysplasia.

Patient and observation

NN, a female new-born of caesarean section delivery at 36 weeks gestation weighted at birth 2700g, her height was 44cm and her head circumference 35 cm. She was born in Lubumbashi in the South of the Democratic Republic of the Congo (D.R.C). Her mother is a 41 years old multiparous, grava 12 para 11. Both her parents are Congolese but non consanguineous and there was no relevant family history.

The clinical examination revealed that NN was born alive and presented a severe hydropsfetalis (Figure 1). Were also noted an important hypoplastic face (Figure 2), a bilateral microtia with ears set low on the head (Figure 3). The new-born presented also a narrow thorax and a protuberant abdomen (Figure 1). Short-limbed dwarfism was observed (Figure 4): short arms and forearms (upper limbs: 11cm) as well as short thighs and legs (lower limbs 14 cm). The hand examination revealed a brachydactyly and a postaxial (ulnar) hexadactyly on all four limbs (Figure 4). The patient died at day 2 before a radiographic examination of the entire skeleton and an abdominal ultra-sound had been performed.

Discussion

Greenberg skeletal dysplasia is an autosomal recessive syndrome extremely rare [3]. This syndrome was described within different ethnic groups and particularly in new-borns with consanguineous parents. Until now, no other case has been reported in the D.R.C, a big country with more than 400 ethnic groups where endogamic marriages are observed only in few of these tribes, the Lunda tribe for example, being one of them. In the case we describe, the female new-born had nonconsanguineous Congolese parents and this is similar to the cases described by Horn et al in 2003 [3] in Germany and by Trajkovski et al [4] in Macedonia. Waterham et al. works, published in 2003 have proved that genetically, Greenberg dysplasia is associated with an inherited disorder of cholesterol biosynthesis caused by LBR gene mutations that lead to the loss of the sterol reductase function of the lamin B receptor [5]. Our patient’s clinical examination revealed a severe hydropsfetalis, a short-limbed dwarfism (upper limbs: 11 cm and lower limbs: 14 cm) and a brachydactyly. Was also observed a postaxial hexadactyly on all four limbs like in the case described by Chitaya et al [6]. A narrow thorax and a protuberant abdomen were observed (Figure 3) as well as a microtia with the ears set low on the head. We were not able to perform different radiographs of the skeleton which might have helped us identify ectopic ossification and/or dysplasial dysplasia as reported in the other cases of Greenberg skeletal dysplasia [1]. An abdominal ultrasound that was not performed either, may have been done to look for an ascites or an intestinal malrotation [3]. This syndrome has a lethal course in most of the reported cases as well as in the first case reported by Greenberg et al. who died in-utero. In our case, the patient was born alive and died two days later.

Conclusion

We have described the first Congolese case of Greenberg skeletal dysplasia, an extremely rare genetic syndrome with only about ten cases to be reported in medical literature up until 2009.

Competing interests

The authors declare no competing interests.

Authors contributions

Toni Kasole Lubala: Redaction of the manuscript, diagnosis, photography.Nina Lubala: Review of the literature and manuscript English translation. Arthur Ndundula Munkana: Management of pregnancy and antenatal diagnosis. Adonis Muganza Nyenga: Correction of the manuscript. Augustin Mulangu Mutombo: Correction of the manuscript. All the authors have read and approved the final version of the manuscript.

Figures

Figure 1: The new-born presented an hydrops fetalis, a narrow thorax and a protuberant abdomen

Figure 2: note the important hypoplastic face

Figure 3: a bilateral microtia with ears set low on the head

Figure 4: Short-limbed dwarfism was observed: short arms and forearms as well as short thighs and legs

References

1. Greenberg CR, Rimoin DL, Gruber HE, DeSa DJ, Reed M, Lachman RS. A new autosomal recessive lethal chondrodysplasty with congenital hydrops. Am J Med Genet. 1988; 29: 623-632. PubMed | Google Scholar

2. Konstantinidou A, Karadimas C, Waterham HR, Supertifurga A, Kaminipetros P, Grigoriadou M et al. Pathologic, radiographic and molecular findings in three fetuses diagnosed with HEM/Greenberg skeletal dysplasia. Prenat Diagn. 2008;28(4): 309-312. PubMed | Google Scholar

3. Horn LC, Faber R, Meiner A, Piskacek U, Spranger J. Greenberg dysplasia: First reported case with additional non skeletal malformations and without consanguinity. Prenat Diagn. 2000; 20(12): 1008-11. PubMed | Google Scholar

4. Trajkovski Z, Vrcakovski, M, Saveski J, and Gucev ZS. Greenberg dysplasia (hydrops-ectopic calcification-moth-eaten skeletal dysplasia): prenatal ultrasound diagnosis and review of literature. Am J Med Genet. 2002; 111 (4): 415-9. PubMed | Google Scholar

5. Waterham HR, Koster J, Mooyer P, NoortGv G, Kelley RI, Wilcox WR, Wanders RJ, Hennekam RC, Oosterwijk JC.
Autosomal Recessive HEM/Greenberg Skeletal Dysplasia Is Caused by 3beta-Hydroxysterol Delta14-Reductase Deficiency Due to Mutations in the Lamin B Receptor Gene. Am J Hum Genet. 2003;72:1013-1017. PubMed | Google Scholar

6. Chitayat D, Gruber H, Mullen BJ, Pauzner D, Costa T, Lachman R, Rimoin DL. Hydrops-ectopic calcification-moth-eaten skeletal dysplasia (Greenberg dysplasia): prenatal diagnosis and further delineation of a rare genetic disorder. Am J Med Genet. 1993;47:272-277. PubMed | Google Scholar

Figure 1: The new-born presented an hydrops fetalis, a narrow thorax and a protuberant abdomen

Figure 2: note the important hypoplastic face
Figure 3: A bilateral microtia with ears set low on the head.

Figure 4: Short-limbed dwarfism was observed: short arms and forearms as well as short thighs and legs.