Meckel–Gruber syndrome in Togo: Prenatal ultrasound and computer tomography diagnosis

**Amadou A., Agbangba K.A., Amoussou K., Douaguibe B.,
Watara G., Sonhaye L., Adjenou V.**

ABSTRACT

Meckel–Gruber syndrome (MGS) is a rare autosomal recessive lethal disorder involving multiple systems. It is characterized by occipital encephalocele, polycystic kidneys and post-axial polydactyly. We report a case of MGS with occipital meningocele, bilateral enlarged echogenic kidneys, polydactyly, and severe oligohydramnios. The diagnosis was made by antenatal ultrasound and computer tomography scan. Meckel–Gruber syndrome is a syndrome rarely described in Africa. Its diagnosis is based on classic triad anomaly. None of this triad seems to be constant.

International Journal of Case Reports and Images (IJCRI)

International Journal of Case Reports and Images (IJCRI) is an international, peer reviewed, monthly, open access, online journal, publishing high-quality, articles in all areas of basic medical sciences and clinical specialties.

Aim of IJCRI is to encourage the publication of new information by providing a platform for reporting of unique, unusual and rare cases which enhance understanding of disease process, its diagnosis, management and clinico-pathologic correlations.

IJCRI publishes Review Articles, Case Series, Case Reports, Case in Images, Clinical Images and Letters to Editor.

Website: www.ijcasereportsandimages.com

CASE REPORT

PEER REVIEWED | OPEN ACCESS

Meckel–Gruber syndrome in Togo: Prenatal ultrasound and computer tomography diagnosis

Amadou A., Agbangba K.A., Amoussou K., Douaguibe B.,
Watara G., Sonhaye L., Adjenou V.

ABSTRACT

Meckel–Gruber syndrome (MGS) is a rare autosomal recessive lethal disorder involving multiple systems. It is characterized by occipital encephalocele, polycystic kidneys and post-axial polydactyly. We report a case of MGS with occipital meningocele, bilateral enlarged echogenic kidneys, polydactyly, and severe oligohydramnios. The diagnosis was made by antenatal ultrasound and computer tomography scan. Meckel–Gruber syndrome is a syndrome rarely described in Africa. Its diagnosis is based on classic triad anomaly. None of this triad seems to be constant.

Keywords: Meckel–Gruber syndrome, Ultrasound, Computer tomography, Togo

How to cite this article

Amadou A., Agbangba K.A., Amoussou K., Douaguibe B., Watara G., Sonhaye L., Adjenou V. Meckel–Gruber syndrome in Togo: Prenatal ultrasound and computer tomography diagnosis. Int J Case Rep Images 2017;8(10):648–652.

Amadou A.¹, Agbangba K.A.¹, Amoussou K.¹, Douaguibe B.², Watara G.¹, Sonhaye L.¹, Adjenou V.¹

Affiliations: ¹Gynecology and Obstetrics Department of CHU Campus; ²Gynécology and Obstétrical Department of CHU Campus.

Corresponding Author: Amadou Abdoulatif, Service de Radiologie et Imagerie, Maître-Assistant, Chu Campus, 05 BP 739 Lomé – Togo.

Received: 02 February 2017

Accepted: 12 June 2017

Published: 01 October 2017

Article ID: Z01201710CR10839AA

doi:10.5348/ijcri-2017100-CR-10839

INTRODUCTION

Meckel-Gruber syndrome (MGS) is a rare and lethal congenital polymorformative syndrome. It is a genetic disease, with autosomal recessive transmission, described for the first time in 1822 by Johan Friedrich Meckel [1]. It is characterized by the triad: encephalocele, cystic dysplasia of the kidneys and polydactyly [2]. Meckel-Gruber syndrome may also cause hepatic developmental defects, and pulmonary hypoplasia. Its global incidence ranges from 1 in 13,250 to 140,000 live births [3]. We report a case of MGS in Togo, whose antenatal diagnosis was made by ultrasound and completed by scanning.

CASE REPORT

A 20-year-old female G1Po presented to the obstetric department for the first time, with history of 28 weeks amenorrhea for routine antenatal examination. There was history of third-degree consanguineous marriage. She had her antenatal examination at a peripheral hospital. She was taking iron and folic acid (dosages of iron and folic acid were normal). Ultrasound done at our hospital as a routine, revealed a fetus with occipital meningocele (Figure 1), bilateral enlarged echogenic kidneys (Figure 2), polydactyly, and severe oligohydramnios. We found no liver abnormalities. For the precise analysis of polydactyly, a CT scan of the uterine contents was made. It showed a postaxial polydactyly affecting the four extremities, with six fingers at each hand and six toes at each foot (Figure 3). The scanner also found an occipital bone defect from which the encephalon herniated.

The family was counseled and after getting consent, termination of pregnancy was planned immediately at 28

weeks' gestation. Post-abortion macroscopic examination revealed multiple congenital anomalies including occipital encephalocele, and post-axial polydactyly.

DISCUSSION

The Meckel–Gruber syndrome, also called dysencephalia splanchnocystica [4], is a rare polymalformative syndrome.

The incidence in Belgium and Finland ranges from 1/3,000–1/9,000, respectively [5]. In India, highest incidence is in Gujrati Indians (1 affected birth per 1,300) [6]. Its incidence is not known in Africa and more particularly in Togo. This is a first discovery in our country. Some cases would probably have gone unnoticed. A high risk (25%) of recurrence in subsequent pregnancies is an important point for consideration and stresses the need

Figure 1: Sonographic intrauterine picture showing defect in the cranium and encephalocele.

Figure 2: Sonographic intrauterine picture showing bilateral dysplastic cystic kidneys.

Figure 3: Computed tomography scan intrauterine image with polydactyly (six digits).

for prenatal diagnosis in expectant mothers in those families. Meckel–Gruber syndrome affects all races with males and females being equally affected [4, 7]. Early, (11–14th week) prenatal ultrasonography is the best method to diagnose MGS [7]. Subsequent autopsy and molecular studies are confirmatory. In our case, ultrasonography found this affection at 28th week.

Meckel–Gruber syndrome is associated with multiple anomalies. It is characterized by classic triad of polycystic dysplastic kidneys, occipital encephalocele (or other anomalies of the central nervous system) and polydactyly [2]. All these three anomalies (triad) were found in our case. According to Sergi et al. [8], polycystic kidneys were found in all cases (100%), occipital encephalocele in 90% of case and post-axial polydactyly in 83.3% of cases. Salonen et al. [5] found a constant association between cystic dysplasia and liver fibrosis, and concluded that these two abnormalities, together with any other abnormalities of the nervous system, suffice to diagnose MGS. Fraser et al. [9] argue that only cystic renal dysplasia is essential for diagnosis. Wrigth et al. [10] never observed cystic dysplasia. The lack of consensus for the diagnosis of MGS led some authors to conclude that none of the anomalies

of the triad or liver fibrosis is essential for diagnosis. The CT scan was almost never used for the diagnosis of MGS in literature. However, it can allow bone analysis in cases where the ultrasound is insufficient and the MRI scan is not available. In our case, the CT scan allowed to diagnose polydactyly and occipital defect.

Meckel–Gruber syndrome has to be differentiated from other syndromes. The most likely syndrome to be confused with MGS is trisomy 13 [3]. Although the dismal outcome is the same for both, the recurrence rate is different. Trisomy 13 is mostly sporadic with low recurrence rate whereas MGS has 25% recurrence rate. Other syndromes similar to MGS are trisomy 18, Joubert syndrome, Bardet–Biedl syndrome and Smith–Lemli–Opitz syndrome [3, 11].

The post-mortem assessment confirms the diagnosis, while genetic studies contribute to the evaluation of the recurrence risk [2, 11].

Given the lethality of MGS in the perinatal or early infantile period, termination of pregnancy should be discussed if available [3].

CONCLUSION

Meckel-Gruber syndrome is a syndrome rarely described in Africa. Its diagnosis is based on classic triad anomaly. None of this triad seems to be constant. The isolation of the gene responsible for this syndrome would therefore be necessary for a diagnosis of certainty.

Author Contributions

Amadou A. – Substantial contribution to conception and design, Analysis and interpretation, Drafting the article, Final approval of version to be published

Agbangba K.A. – Substantial contribution to conception and design, Analysis and interpretation, Drafting the article, Final approval of version to be published

Amoussou K. – Substantial contribution to conception and design, Analysis and interpretation, Drafting the article, Final approval of version to be published

Douaguibe B. – Substantial contribution to conception and design, Analysis and interpretation, Drafting the article, Final approval of version to be published

Watara G. – Substantial contribution to conception and design, Analysis and interpretation, Drafting the article, Final approval of version to be published

Sonhay L. – Substantial contribution to conception and design, Analysis and interpretation, Drafting the article, Final approval of version to be published

Adjenou V. – Substantial contribution to conception and design, Analysis and interpretation, Drafting the article, Final approval of version to be published

Guarantor

The corresponding author is the guarantor of submission.

Conflict of Interest

Authors declare no conflict of interest.

Copyright

© 2017 Amadou A. et al. This article is distributed under the terms of Creative Commons Attribution License which permits unrestricted use, distribution and reproduction in any medium provided the original author(s) and original publisher are properly credited. Please see the copyright policy on the journal website for more information.

REFERENCES

1. Meckel JR. Beschreibung zweier durch sehr ähnliche Bildungsabweichung entsellter Geschwister [Article in Jarman]. *Dtsch Arch Philol* 1898;7:99.
2. Paavola P, Salonen R, Baumer A, et al. Clinical and genetic heterogeneity in Meckel syndrome. *Hum Genet* 1997 Nov;101(1):88–92.
3. Alexiev BA, Lin X, Sun CC, Brenner DS. Meckel-Gruber syndrome: Pathologic manifestations, minimal diagnostic criteria, and differential diagnosis. *Arch Pathol Lab Med* 2006 Aug;130(8):1236–8.
4. Salonen R, Paavola P. Meckel syndrome. *J Med Genet* 1998 Jun;35(6):497–501.
5. Salonen R, Norio R. The Meckel syndrome in Finland: Epidemiologic and genetic aspects. *Am J Med Genet* 1984 Aug;18(4):691–8.
6. Young ID, Rickett AB, Clarke M. High incidence of Meckel's syndrome in Gujarati Indians. *J Med Genet* 1985 Aug;22(4):301–4.
7. Naveen NS, Vishal K, Vinay KV. Meckel Gruber Syndrome: A case report. *Nitte University Journal of Health Science* 2013;3:83–5.
8. Sergi C, Adam S, Kahl P, Otto HF. Study of the malformation of ductal plate of the liver in Meckel syndrome and review of other syndromes presenting with this anomaly. *Pediatr Dev Pathol* 2000 Nov–Dec;3(6):568–83.
9. Fraser FC, Lytwyn A. Spectrum of anomalies in the Meckel syndrome, or: “Maybe there is a malformation syndrome with at least one constant anomaly”. *Am J Med Genet* 1981;9(1):67–73.
10. Wright C, Healicon R, English C, Burn J. Meckel syndrome: What are the minimum diagnostic criteria? *J Med Genet* 1994 Jun;31(6):482–5.
11. Chen CP. Meckel syndrome: Genetics, perinatal findings, and differential diagnosis. *Taiwan J Obstet Gynecol* 2007 Mar;46(1):9–14.

ABOUT THE AUTHORS

Article citation: Amadou A., Agbangba K.A., Amoussou K., Douaguibe B., Watara G., Sonhay L., Adjenou V. Meckel–Gruber syndrome in Togo: Prenatal ultrasound and computer tomography diagnosis. Int J Case Rep Images 2017;8(10):648–652.

Abdoulatif Amadou is resident at Department of Radiology, ICHU Campus, University of Lome, Lome, Togo. His research interests include neuroradiology and obstetrical ultrasound.
Email: amadoulatif@yahoo.fr

Agbangba K.A., Radiology Department of CHU Campus.

Amoussou K., Radiology Department of CHU Campus.

Douaguibe B., Gynecology and Obstetrics Department of CHU Campus.

Watara G., Radiology Department of CHU Campus.

Sonhay L., Radiology Department of CHU Campus.

Adjenou V., Radiology Department of CHU Campus.

Access full text article on
other devices

Access PDF of article on
other devices

Edorium Journals: An introduction

About Edorium Journals

Edorium Journals is a publisher of international, high-quality, open access, scholarly journals covering subjects in basic sciences and clinical specialties and subspecialties.

Invitation for article submission

We sincerely invite you to submit your valuable research for publication to Edorium Journals.

Why should you publish with Edorium Journals?

In less than 10 words: "We give you what no one does".

Vision of being the best

We have the vision of making our journals the best and the most authoritative journals in their respective specialties. We are working towards this goal every day.

Exceptional services

We care for you, your work and your time. Our efficient, personalized and courteous services are a testimony to this.

Editorial review

All manuscripts submitted to Edorium Journals undergo pre-processing review followed by multiple rounds of stringent editorial reviews.

Peer review

All manuscripts submitted to Edorium Journals undergo anonymous, double-blind, external peer review.

Early view version

Early View version of your manuscript will be published in the journal within 72 hours of final acceptance.

Manuscript status

From submission to publication of your article you will get regular updates about status of your manuscripts.

Our Commitment

Six weeks

We give you our commitment that you will get first decision on your manuscript within six weeks (42 days) of submission. If we fail to honor this commitment by even one day, we will give you a 75% Discount Voucher for your next manuscript.

Four weeks

We give you our commitment that after we receive your page proofs, your manuscript will be published in the journal within 14 days (2 weeks). If we fail to honor this commitment by even one day, we will give you a 75% Discount Voucher for your next manuscript.

Favored author program

One email is all it takes to become our favored author. You will not only get 15% off on all manuscript but also get information and insights about scholarly publishing.

Institutional membership program

Join our Institutional Memberships program and help scholars from your institute make their research accessible to all and save thousands of dollars in publication fees.

Our presence

We have high quality, attractive and easy to read publication format. Our websites are very user friendly and enable you to use the services easily with no hassle.

Something more...

We request you to have a look at our website to know more about us and our services. Please visit:
www.edoriumjournals.com

We welcome you to interact with us, share with us, join us and of course publish with us.

Edorium Journals: On Web

Browse Journals

CONNECT WITH US