NIGERIAN JOURNAL OF FAMILY PRACTICE
*The official publication of the Society of Family physicians of Nigeria*

**Vol. 7. Number 1. April. 2016 ISSN: 2141 - 9884**

# TABLE OF CONTENTS

# JUVENILE POLYPOSIS COLI IN A 9 YEAR OLD BOY

**Edwin Oseni-Momodu[1], Chima AA George[2], Pius Ameh[2], Adam Lee[1], Paul Ushie[2], Lengmang Sunday[2], Judy HO[1],**

[1]Department of Surgery, Bingham University/Bingham University Teaching Hospital.
[2]Department of Family Medicine, Bingham University/Bingham University Teaching Hospital.
Corresponding Author: Edwin Oseni-Momodu.

**Key words:** Juvenile polyposis syndrome, Juvenile polyposis coli, colo-rectum, Polyps, auto-amputation

**Conflict of interest:** We declare that we have no financial or personal relationship which may have inappropriately influenced us in writing this paper.

## ABSTRACT

Juvenile Polyposis syndrome (JPS) is a rare disease condition characterised by development of multiple polyps commoner in the colo – rectum. An autosomal dominant condition usually associated with mutations in 2 genes – BMPRIA (bone morphogenic protein receptor, type I A) and SMAD4 (mothers against decapentaplegic, drosophilia, homolog of 4).

The authors present the case of a nine year old male from a poor family in Kano, North West Nigeria but residing in Jos, whose parents were terrified by his frequent passage of bloody stool and on one occasion passed a strange object in the stool that turned out to be Juvenile polyposis coli.

Through collaborative effort of the authors and their partner in Hong Kong, screened the entire family and sent samples to Hong Kong for further gene analysis. The gene analysis is expected to reveal a genetic background of an autosomal dominant genetic disorder that will justify the screening of the entire family for Juvenile Polyposis Syndrome. Such information is necessary to start Familial Polyposis Coli register.

No case of JPS has been reported in North Central, Nigeria where the authors practice, this being the very first from ECWA Evangel Hospital now Bingham University Teaching Hospital; since its inception in 1959.

## INTRODUCTION

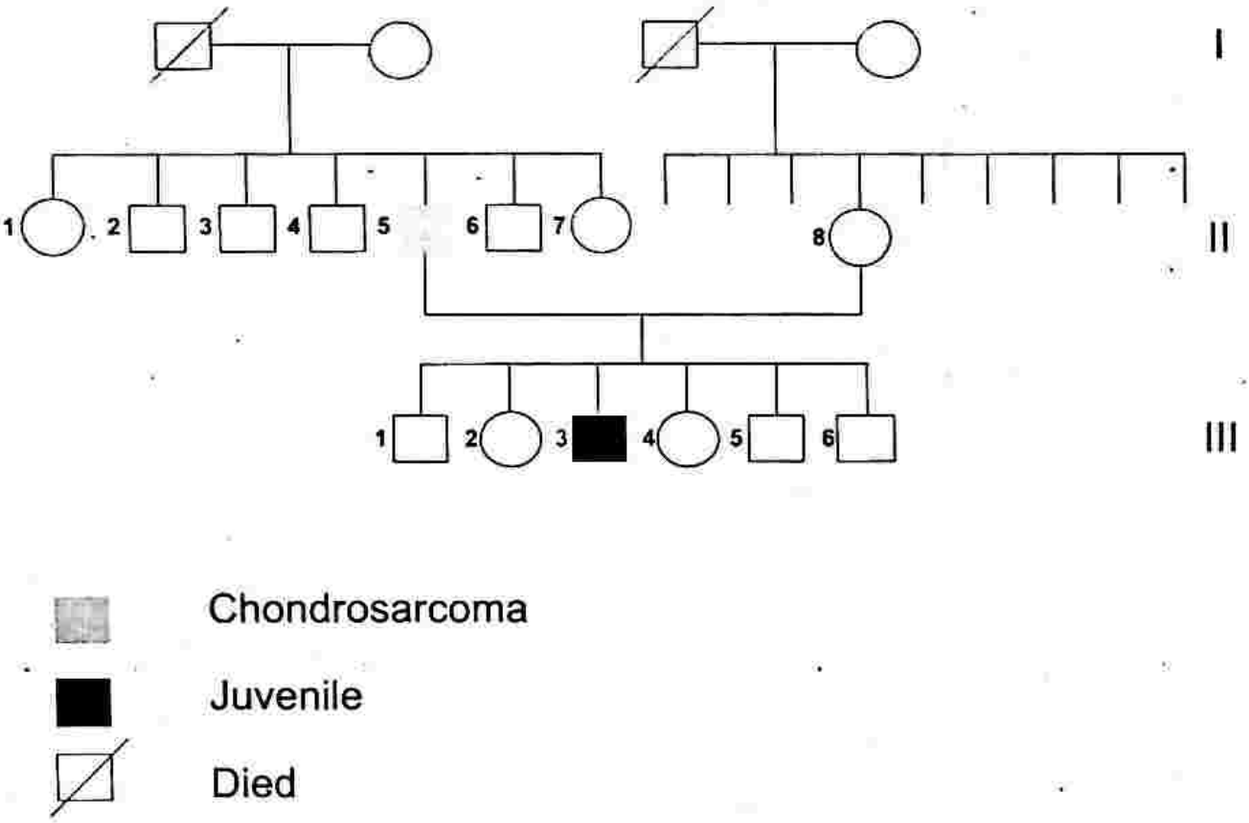
Juvenile polyposis syndrome is a rare disease entity that is characterized by multiple non – cancerous or benign growths otherwise called juvenile polyps.[1, 2, 3] Generally gastrointestinal polyps are common cause of rectal bleeding in toddlers and pre-school age children.[4] They are harmatomatous lesions occurring in gastrointestinal tract characterised by smooth histological appearance, predominant stroma, cystic spaces and lack of a smooth muscle core.[5] Majority of the polyps in children are juvenile in nature and mostly located in the colorectal region.[4]

In a review of 563 children aged between 2 months and 17 years, the highest incidence occurred in the age range between 2 and 10 years.[4] These group of the population represented about 85 per cent of the study population. In that review the patients presented with rectal bleeding in 78.5 per cent of the cases. Although most of the juvenile polyps are benign, there is however risk of progression to malignancy when missed thus calling for diligent follow up and management of each case diagnosed. Juvenile polyps are existent in a number of mendelian disorders, occasionally in association with gastrointestinal cancers (Juvenile polyposis syndrome) and at some other times as part of known syndromes like Cowden Gorlin and Banayan – Zonana in association with growing anormalies.[5,6] The prevalence ranges between 1 in 16, 000 to 1 in 100,000 individuals.[7,8] Although most juvenile polyps are non-cancerous but there are 10 to 50 per cent chances of transformation into a cancer.[8,9] The gravest complication that occurs later on in life is cancerous transformation and the average age of cancer development is about 35 years of age.[7,8,9] The most commonly occurring cancer in people with juvenile polyposis syndrome is colorectal cancer.[7,8,9]

The polyps developed can lead to gastrointestinal bleeding which in-turn leads to reduced number of red blood cells or anaemia, abdominal pain and diarrhoea. About 15 per cent cases of juvenile polyposis do manifest other anomalies like intestinal mal-rotation in which the intestine is twisted, heart or brain anomalies, cleft palate, extra fingers or toes, also called polydactyl and anomalies of the genitalia or urinary tract.

In this case report, we present a 9 year old male whose parents brought an auto-amputated polyp that the boy passed out while daefecating, wandering what it was that their child had passed out. In this particular case further examinations and investigations confirmed it to be juvenile polyposis coli.

Figure 1




Generation 1, 1-died of hypertension, 2 – 70 years of age and well, 3 – died? age, 4 – Alive and well; Generation 2, 1 – DOB 1956, suffered from diabetes mellitus and poliomyelitis, children are well, 2 – DOB, 1960; well, children are well; 3 – DOB, 1963, well, children well, 4

**CASE REPORT**

A 9 year old male Hausa boy presented to our surgical outpatient clinic in early August; 2013 with rectal bleeding and passage of a polyp during daefecation. This was a surprise to the parents who retrieved the polyp from the bloody stool and subsequently brought it with them alongside the boy to the clinic. The polyp was subsequently preserved in formalin and sent to the pathologist for analysis. The result later came out to be a polyp. Besides the bleeding per rectum and passage of a polyp, he also had a history of intermittent left upper quadrant abdominal pain. The physical examination showed that he was generally in good condition and well nourished. There was no dysmorphic features and he was of normal intelligence.

The abdominal examination revealed a soft, non – tender abdomen with no distension and no mass was palpable. The digital rectal examination was normal.

Upper endoscopy carried out on the 15th of August; 2013 showed a normal oesophagus, gastrum and duodenum up to the first part of the duodenum.

A flexible sigmoidoscopy up to 50cm above the ano – cutaneous verge carried out on the same 15th of August; 2013 revealed five non – bleeding polyps that varied between 2mm and 5mm in size in the descending colon and rectum with no apparent malignant change. Attempt at colonoscopy of the boy was aborted as a result of instrument failure.

Blood sampleswere collected, preserved and sent to our partners in Hong Kong for gene analysis and we are presently awaiting the result as we continue to follow up the entire family and in view of this have drawn the family genogram as shown in figure 1 below:

**DISCUSSION**

Juvenile polyposis syndrome is a condition, characterised by multiple harmatomatous polyps that in 98 per cent of the cases invade the gastrointestinal tract, colon and rectum, the gastrum in 13.6 per cent, duodenum in 2.3 per cent, jejunum and ileum in 6 per cent of the cases.[9] Most of the polyps described above occur in the colonic region and although colonic polyps are said to be rare in the Africans, recent reports suggest a rising incidence of colonic polyps among Africans.[10] Juvenile polyposis syndrome can be transmitted from one individual to another by autosomal dominant inheritance and has cancer predisposition tendency.[11]

The syndrome is characterised by the development of harmatomatous polyps throughout the gastrointestinal tract that includes the small intestine, colon and rectum. The term" juvenile" does not refer to the age of onset in the individual affected but more to the specific type of polyp found. The histology usually give a picture of a normal epithelium with dense stroma, inflammatory infiltrate and a smooth surface with dilated mucus - filled cystic glands in the lamina propria.[1] In terms of manifestation majority of the juvenile polyps are benign and often lead to clinically significant bleeding and malignant transformation in some cases. The life time risk of developing cancer is said to be between 9 per cent to 50 per cent and of those that eventually transform into a cancer colorectal carcinoma is in the majority.[8,11]

Clinically the diagnosis of juvenile polyposis syndrome can be made when one of the features below is met:[3,6,11]

1. If there are more than five juvenile polyps in the colorectum
2. If there are multiple juvenile polyps throughout the gastrointestinal tract.
3. Any number of juvenile polyps and a family history of juvenile polyps

The index patient presented in this case report qualifies as a case of juvenile polyposis syndrome since one polyp was already passed out during daefecation while five were seen during endoscopic investigation.

There are three types of juvenile polyposis syndrome described on the basis of the signs and symptoms they present with; and these include:[7,9]

1. Juvenile polyposis of infancy (JPI) is the most severe form of these entities and also has the worst outcome. The characteristics of JPI include the occurrence of polyps throughout the gastrointestinal tract during infancy. Children with this type of polyposis may develop protein – losing enteropathy characterised by severe diarrhoea, lack of weight gain and expected growth rate or failure to thrive; generalised wasting and weight loss or cachexia.
2. The second type is that known as generalized juvenile polyposis diagnosed when polyps are found throughout the gastrointestinal tract.
3. The third is known as juvenile polyposis coli in which the polyps are found only in the affected individual's colon.

The last two conditions described under types of juvenile polyposis usually develop polyps during their childhood. Although juvenile polyps are rare they are however common in the causation of rectal bleeding in children and should be suspected in case of haematochezia in children especially when recurrent.[11,12] Our patient can be said to belong to the third category and he presented with history of rectal bleeding as well as abdominal pain which is also a common symptom of juvenile polyposis syndrome.

The pattern of inheritance of juvenile polyposis syndrome is by autosomal dominant fashion in which case a copy of the altered gene in each cell is enough for the disease condition to manifest.[7] In about 75 per cent of cases the affected person inherits the mutant gene from one affected parent while the rest 25 per cent occur as a result of new mutations in the gene and will manifest in people without prior history of the disorder in their household.[13]

The aetiology of JPS can be due to mutations in two genes namely the SMAD4/DPC4 that is located on chromosome 18q21 and the BMPR1A located on chromosome 10q21 – 22. The genetic testing for JPS is indicated by the presence of three to five juvenile polyps in a patient, the detection of a juvenile polyp and a family history of JPS, or being a parent or sibling of one known to be a carrier of mutation.[13] For confirmation of JPS on a molecular level the patient will undergo gene testing that includes use of the patient's blood or saliva sample obtained. The individual's DNA is isolated from the sample and two copies of BMPR1A and SMAD4. Once a mutation in either of these two genes is identified, the genetic counsellor should then examine if the alteration has been previously reported in other individuals with juvenile polyposis syndrome.

The genetic testing for mutations in SMAD4 and BMPR1A involves DNA sequencing and in 35 to 50 per cent of patients there is association with JPS.[13]

The differential diagnosis of juvenile polyposis syndrome include Cowden disease, PeutzJeghers syndrome, conkhite – Canada syndrome, Hereditary mixed polyposis syndrome, Gastric Hyperplastic polyp, Serrated (Hyperplastic) polyposis and Familiar adenomatous polyposis.[5,8]

There are no medicines for the treatment of juvenile polyposis syndrome however it is important that the clinician pay close attention to the patient by way of screening with continuous monitoring and follow up of the patient and the entire family. The screening and continuous monitoring include blood test, colonoscopy and upper endoscopy which should be carried out when the patient with JPS is 15 or when

symptoms first appears.[13,14] If results are negative then the person should be screened three years after. When only a few polyps are found the polyps should be removed at endoscopy and screening carried out yearly until no polyps are found; then subsequent screening should be every three years. If the patient requires surgery then screening should be every year until no polyps are found and then every three years.[14] Prophylactic polypectomy can be done during endoscopy for few polyps while in severe cases varying degrees of colectomy or gastrectomy are carried out. This also helps in preventing cancer as a complication of JPS.

The prognosis of JPS varies. There is 9 to 50 per cent risk of developing of the gastrointestinal tract.[14]This is commoner in those whose polyps occur in the colon, though it can also occur in the small intestine, stomach or pancreas. To this end it is very important to promptly screen and remove the polyps on presentation. Strong link exists between JPS and hereditary haemorrhagic telangiectasia (HHT) in persons with SMAD4 mutation and where the JPS is due to an SMAD4 mutation, an additional HHT related tests and screening are indicated.[14]

For the patient he simply wanted to recover from whatever was causing the rectal bleeding but for the parents of the patient they wanted to know why their child should pass such strange object through the anus and what the strange object that the child had passed was and hoped that such would not occur again since they were visibly afraid of what the outcome might be.

The authors in writing this case have drawn a lesson in collaborative effort even at international level; at patient care as they were able to muster support for the patient that included sending blood samples for gene analysis and on-going support for follow up of the patient since the entire family were too poor to pay their hospital bills.

## CONCLUSION

Juvenile polyposis syndrome is not a common disease. There is a definite possibility of transformation to malignancy and thus there is a need for prophylactic surgery. The patients also need to be followed up regularly as well as regular and active surveillance of the family members. In the case above a multidisciplinary approach involving 2 general surgeons, a colorectal surgeon and family physicians were involved and still involved in the management of the patient and surveillance of the entire family.

## REFERENCES:


1. Mounia El Yousfi1, Bahija Benyachou, Adil Ibrahimi1, Laila Chbani, AfafAmarti. Young Patient with Juvenile Polyposis Syndrome: A case report. Open Journal of Gastroenterology 2 (2012) 105-108.
2. Joel Johansson, ChristoferSahin, Rebecka Pestoff, Simone Ignatova, Pia Forsberg, Anders Edsjö, et al. A Novel SMAD4 Mutation Causing Severe Juvenile Polyposis Syndrome with Protein Losing Enteropathy, Immunodeficiency, and Hereditary Haemorrhagic Telangiectasia; Hindawi Publishing Corporation Case Reports in Gastrointestinal Medicine, Volume 2015, 1 – 5 accessed online on the 21'' of August; 2015 at http://dx.doi.org/10.1155/2015/140616.
3. Steven Kelly, Simon Dwerryhouse, Peter Safranek , Richard Hardwick. Juvenile polyposis syndrome affecting the stomach: a case report. *Journal of Medical Case Reports* 2008, 2:314 doi: 10.1186/1752-1947-2-314.
4. Sreekant Avula, Bhavuray Teli, Satish Arakeri, Venugopal PR. Prolapsed rectal polyp in an infant with massive lower gastrointestinal bleeding: case report, *International Surgery Journal*, 2015 May;2(2):320-322.
5. Richard Houlston, Stephen Bevan, Andrew Williams, Joanne Young, Malcolm Dunlop, Paul Rozen et al. Mutations in DPC4 (SMAD4) cause Juvenile Polyposis Syndrome, but only account for a minority of cases.
6. Anne Marie Jelsig, NielsQvist, Klaus Brusgaard, Claus Buhl Nielsen, Tine Plato Hansen, Lilian Bomme Ousager. Hamartomatous polyposis syndromes: A review; Jelsig et al. *Orphanet Journal of Rare Diseases* 2014, 9:101 http://www.ojrd.com/content/9/1/101
7. Mikoaj Teisseyre, Andrzej Awski, Marta Podralska, RyszardSomski,Hor Ismail, Marek Woynarowski et al. Morphological features of juvenile polyposis syndrome associated with new detected BMPR1A gene mutation. Case report. *Annals of Diagnostic Paediatric Pathology* 2007, 11(3–4):115–118.
8. Nitin Mishra, Jason Hall. Identification of Patients at Risk for Hereditary Colorectal Cancer; Clinics in Colon and Rectal

Surgery, 2012 Vol. 25 Num. 2, Pg. 67-82.

9. Beatrix Tam, Ágnes Salamon, Attila Bajtai, Annamária Németh, János Kiss, László Simon et al. The real face of juvenile polyposis syndrome. *J Gastrointest Oncol* 2012;3(4):362-368.
10. OI Alatise, AO Arigbabu, AE Agbakwuru, OO Lawal, OA Sowande, OO Odujoko et al. Polyp prevalence at colonoscopy among Nigerians: A prospective observational study. *Nigerian Journal of Clinical Practice*; Nov-Dec 2014, Vol 17(6), pg756-762.
11. Zsofia K. Stadler, Erin Salo-Mullen, Liying Zhang, Jinru Shia, Ruben Bacares, Derek G. Power et al. Juvenile Polyposis Syndrome Presenting With Familial Gastric Cancer and Massive Gastric Polyposis. *Journal of Clinical Oncology*; September 1 2012. Vol. 30, Num. 25 pg. c229-c232.
12. Anyanwu Lofty-John C, Mohammad Aminu M, Ibrahim Mustapha U, Atanda Akinfenwa T. Juvenile polyp in children in Kano Nigeria: Clinical presentation and management challenges. *Nigerian Journal of Basic and Clinical Sciences* / Jan-Jun 2014 / Vol 11 | Number 1 pg. 20–23.
13. Children's Hospital of Philadelphia, Juvenile Polyposis Syndrome, 2015; Accessed online on the 11th of September; 2015 at http://www.chop.edu/conditions-diseases/juvenile-polyposis-syndrome#.VfLMv_RcBmk
14. Cleveland Clinic Foundation, Juvenile Polyposis Syndrome; 2013. Accessed online on the 11th of September; 2015 at http://my.clevelandclinic.org/childrens-hospital/health-info/diseases-conditions/hic-juvenile-polyposis-syndrome