Oral-Facial-Digital Syndrome with Hirschsprung Disease - A New horizon

Roopak Dubey¹, Kamal Kumar Sen², Rudra Narayan Dash³, Mayank Goyal⁴

¹1st Year Post Graduate Trainee, ²Professor and HOD, ³Associate Professor, ¹1st Year Post Graduate Trainee, Department of Radio-diagnosis, Kalinga Institute of Medical Sciences, Bhubaneswar, Odisha, India

Corresponding author: Dr. Roopak dubey, Department of Radio-diagnosis, Kalinga Institute of Medical Sciences, KIIT Road, Patia, 751024, Bhubaneswar, India

OdishaDOI: http://dx.doi.org/10.21276/ijcmsr.2020.5.1.34


ABSTRACT

Introduction: Oral-facial-digital syndrome (OFDS) is a rare disorder and its association with Hirschsprung disease (HSCR) makes it further more infrequent. The main aim and objective of this paper is to enhance the understanding between Hirschsprung disease and Oral facial digital syndrome.

Case report: We are presenting here a 8 months old baby boy with biopsy proven Hirschsprung disease along with morphological anomalies consistent with Oral-Facial-Digital syndrome. A previously undescribed case of Hirschsprung disease with post axial polydactyly, hypertelorism, ASD, hyperplastic frenulum, high arched palate, depressed nasal bridge, low set ears and frontal bossing is presented here.

Conclusion: The presented case showed some similarities to “Unclassified variant” of OFDS but there were some differences also which had not been described in the literature earlier. Hence, this case can be considered either as an extended version of “Unclassified variant” of OFDS or a new variant of OFDS.

Keywords: Oral-facial-digital Syndrome, Hirschsprung Disease, Variant

INTRODUCTION

Oral-facial-digital syndrome (OFDs) is a group of rare disorders characterised by abnormalities of oral cavity, face and digits of hands and feet¹. Its association with Hirschsprung disease makes it further uncommon. The relationship of ciliopathies like Oral-facial-digital syndrome, Joubert syndrome and Meckel Gruber Syndrome with neurocristopathy like Hirschsprung disease (HSCR) has been described in the literature, however there is lack of clear understanding between their association and finding of a common genetic/molecular associating link between the two is under progress. A rare association of Hirschsprung disease with probably an extended version of unclassified variant of OFDs or a new variant of OFDs is presented in this report.

CASE REPORT

A 8 months old very irritable male baby from non-consanguineous parents, was brought to our Institution with bilious vomiting and abdominal distention for 12 hours. His mother stated that the infant had been constipated since birth and failed to pass meconium during the first 48 hours of life. Distended bowel loops were noted on X-ray (Fig.1). A subsequent rectal biopsy revealed absence of ganglionic cells in colonic wall, confirming the diagnosis of Hirschsprung disease. He was treated surgically by “Duhamel’s retrorectal pull through” procedure. There was no family history of congenital anomalies. Besides Hirschsprung disease, several morphological congenital defects were also noted. Left hand showed post-axial polydactyly where two digits arising from 4th metacarpal (Fig.2A). In addition there was Y shaped 3rd and small under-developed 4th metacarpal with post-axial polydactyly in right hand (Fig.2B). Left foot showed Y shaped 4th metatarsal and post-axial polydactyly (Fig.2C). So, digital features include post-axial polydactyly in all the limbs except right foot.

Figure-1: Pre-operative radiograph of abdomen showing distended bowel loops.
正常男性染色体模式为46 XY。

**DISCUSSION**

Hirschsprung病呈现出与相关染色体异常12%的病例和额外的先天性异常18%的病例。常见相关先天性异常与Hirschsprung病包括腭裂、多指(趾)、心脏隔膜缺陷、消化道畸形和颅面异常。3. 迄今为止记录的变体有：
1. 肛管直肠畸形、单侧肾缺如、前额突出和先天性耳聋。
2. 肛管直肠畸形、后前肢多指和室间隔缺损。
3. 肛管直肠畸形、远端指骨和皮肤的轻度异常。
4. 肛管直肠畸形、心脏缺陷和喉部异常。
5. 肛管直肠畸形、并指、脑室发育不良和脊柱异常。
6. 肛管直肠畸形、并指。

据我们所知，目前的情况与先前未描述的变体的“未分类的变体”有关。在临床表现中，我们观察到近端多指(趾)、融合肾脏、TOF/VSD、舌体发育不良、肢体缺失、心脏发育不良和智力障碍。与未分类的变体中融合肾脏的差异在于正常USG检查未显示任何肾脏异常。ASD在我们的病例中被发现，与TOF/VSD、未分类的变体中室间隔缺损的组合不同。此外，与融合肾脏和融合舌体通常在未分类的变体中出现相反，我们的患者有增生的系带和高腭弓。不同变体最相关的特征如表1所示。

**CONCLUSION**

根据我们的患者，显然它并不符合OFD的任何一种类型。最接近的亚型是未分类的变体。因此，我们相信这可能是未分类变体或OFD中未描述的变体的变体。
positive family history in this case implies that the OFD malformations could have resulted from a spontaneous mutation, an X-linked dominant trait or an autosomal recessive trait with minimal expression in the preceding generations.

REFERENCES

Source of Support: Nil; Conflict of Interest: None
Submitted: 29-11-2019; Accepted: 20-12-2019; Published online: 28-02-2020