Case 15381

Eurorad

Oro facial digital syndrome type 1 with heterotopia, polymicrogyria and corpus callosum agenesis - a rare case report

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Section: Abdominal imaging
Area of Interest: Ear / Nose / Throat Musculoskeletal system Kidney Pancreas Biliary Tract / Gallbladder Head and neck
Procedure: Observer performance
Procedure: Computer Applications-General
Procedure: Imaging sequences
Imaging Technique: Experimental
Imaging Technique: Ultrasound
Imaging Technique: MR
Special Focus: Congenital Cysts Dilatation Case Type: Clinical Cases
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Patient: 28 years, female

Clinical History:

A 28-year-old female presented with epigastric pain. On general examination brachydactyly was seen in hands and feet. Mild cognitive impairment with lobulated tongue, abnormal dentition and patchy areas of baldness were other associated findings.

Imaging Findings:

Ultrasonography of the entire abdomen was done and cholelithiasis was found to be the cause of pain. In addition, bilateral enlarged polycystic kidneys with multiple cysts in pancreas and beaded dilatation of intrahepatic biliary radicles was also seen. Magnetic resonance cholangiopancreatography was done for further evaluation which did not add any other findings. MRI brain was also done, which revealed heterotopia, polymicrogyria, complete corpus callosum agenesis and bilateral frontal lobe atrophy.

Discussion:

Oro-facial-digital syndrome (OFDS) is a group of various genetic disorders which cause malformations of the face, hands, and feet.
There are many known subtypes of OFDS which include:[1]

OFDS type I: Gorlin-Psaume syndrome/Papillon-Leage syndrome
OFDS type II: Mohr syndrome/Mohr-Claussen syndrome
OFDS type III: Sugarman syndrome
OFDS type IV: Baraitser-Burn syndrome
OFDS type V: Thurston syndrome
OFDS type VI: Juberg-Hayward syndrome/Varadi-Papp syndrome
OFDS type VII: Whelan syndrome
OFDS type VIII: Edwards syndrome
OFDS type IX: OFDS with retinal abnormalities
OFDS type X: OFDS with Fibular aplasia

OFDS are associated with malformations of-
*Face (facial asymmetry, hypoplastic mandible, hypertelorism)
*Oral cavity (cleft lip and palate, lobulated tongue, abnormal dentition)
*Digits (brachydactyly, clinodactyly, syndactyly, polydactyly)
In addition cysts in liver, pancreas and central nervous system anomalies such as agenesis of corpus callosum, cortical hypoplasia, ventricular asymmetry, polymicrogyria and heterotopic grey matter are also seen.
Polycystic kidney disease is one of the major diagnostic determinants of OFDS type 1.[2]
Genetic work-up for the detection of mutated OFD1 gene or development of polycystic kidney disease in adulthood confirms the diagnosis of OFD1. Treatment includes correction of craniofacial, digital abnormaliities surgically and counselling to cope with learning disabilities. [3] The treatment targets on improving the patient's confidence to live a near normal life.

Differential Diagnosis List: Oro- facial- digital syndrome type 1, Oro facial digital syndrome type VI, Oro facial digital syndrome type III

Final Diagnosis: Oro- facial- digital syndrome type 1

References:

Franco B, Thauvin-Robinet C (2016) Update on oral-facial-digital syndromes (OFDS). cilia 5: 12 (PMID:27141300)
Khalifa, Ola; Rahbeeni, Zuhair; Alhashmi, Nadia; Almane, Khalid (2012) Oral–facial–digital syndrome type 1: unique radiological findings. Clinical Dysmorphology 21;71-79 (PMID: 22123492)
Connacher AA, Forsyth CC, Stewart WK (1987) Orofaciodigital syndrome type I associated with polycystic kidneys and agenesis of the corpus callosum. Journal of Medical Genetics 24:116-118. (PMID:3560170)

Figure 1

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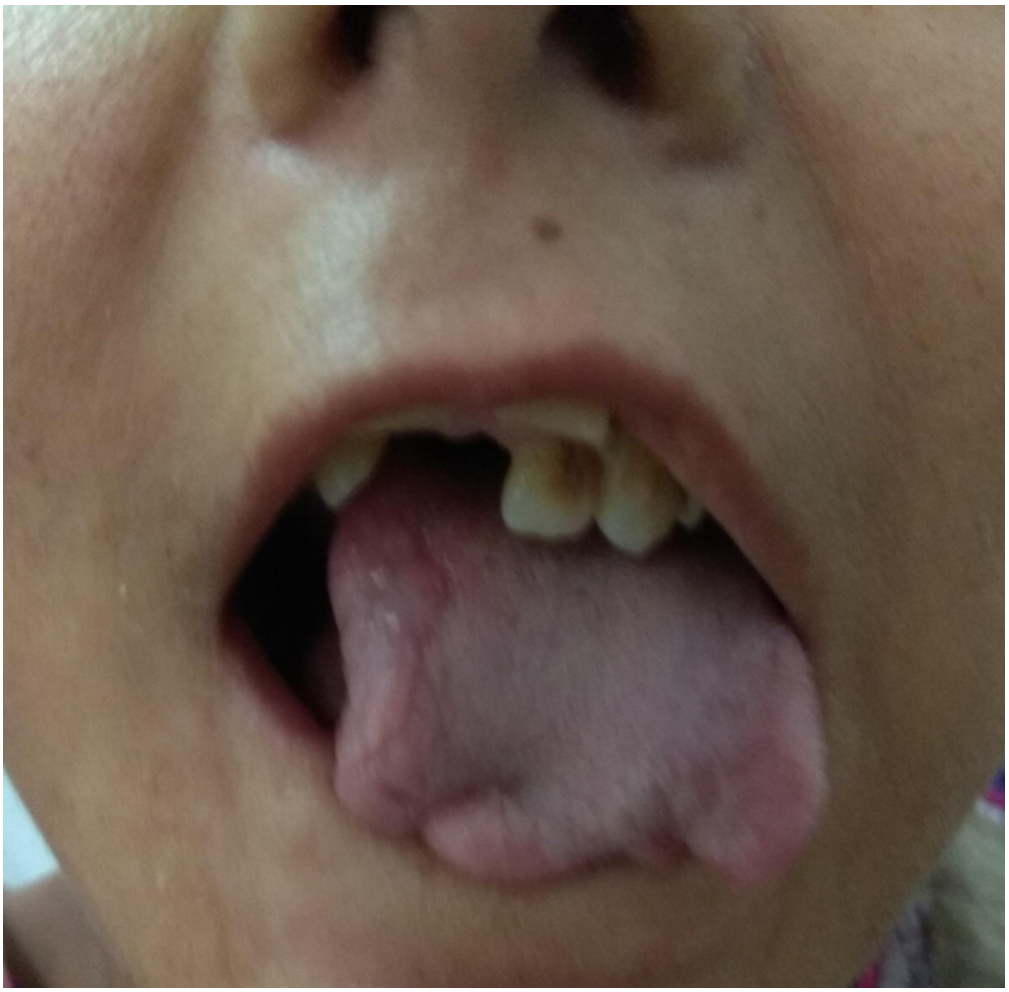
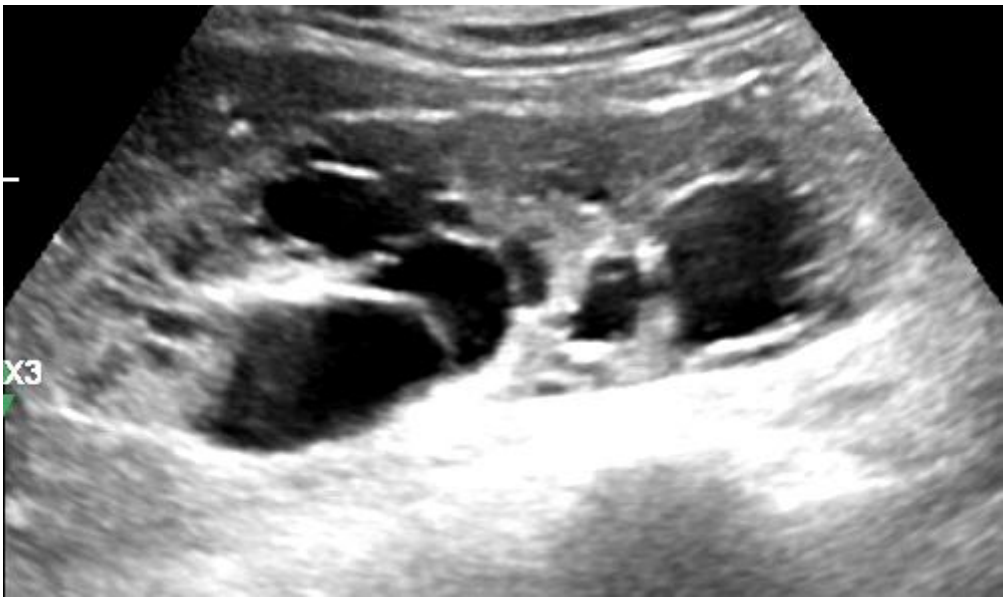


Description: Oral examination showing lobulated tongue and abnormal dentition. **Origin:** Arjumand, Department of Radiodiagnosis, King George's Medical University, Lucknow, India

Figure 2

a



Description: Sonography showing multiple cysts of varying sizes in kidneys- polycystic kidneys. **Origin:** Arjumand, Department of Radiodiagnosis, King George's Medical University, Lucknow, India

Figure 3

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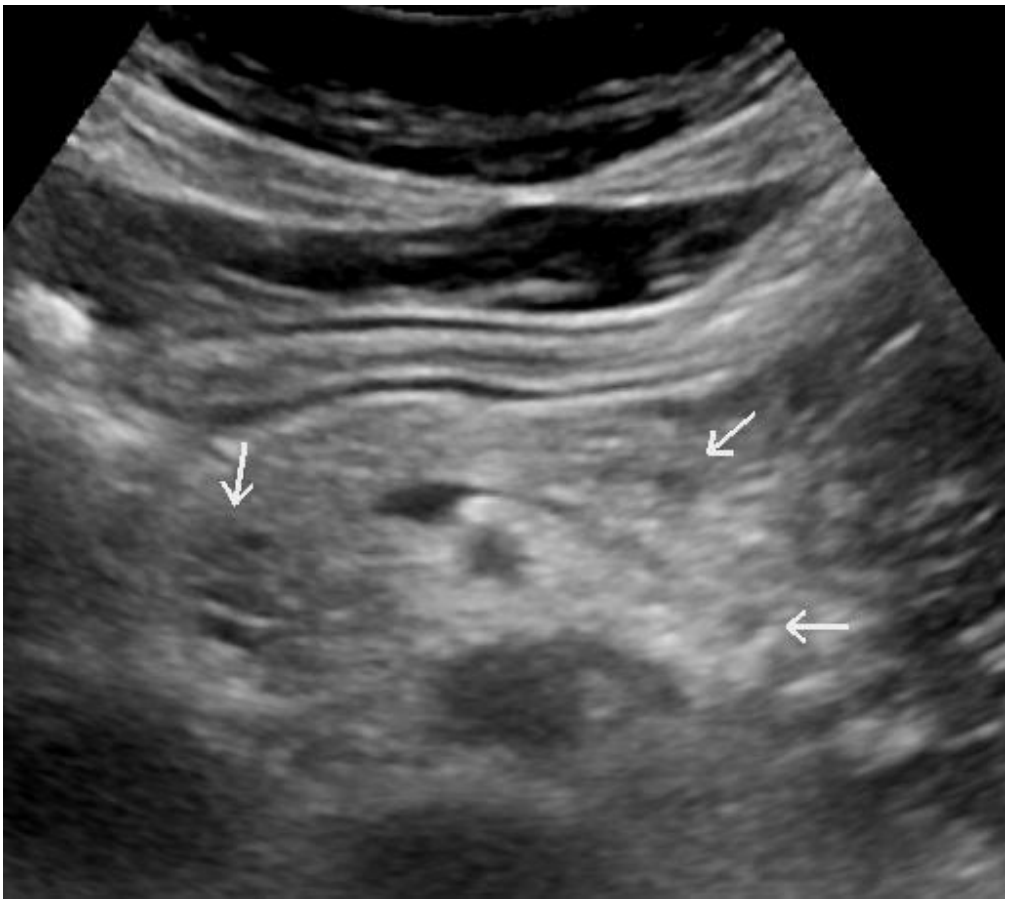

Description: Sonography showing few tiny cysts in pancreatic parenchyma. **Origin:** Arjumand, Department of Radiodiagnosis, King George's Medical University, Lucknow, India

Figure 4

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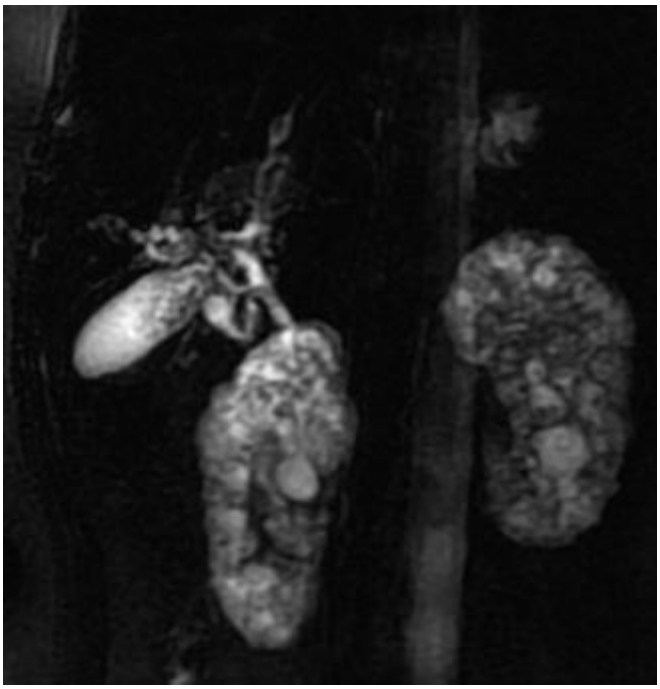

Description: Thick slab MRCP images showing beaded dilatation of intrahepatic biliary radicles and bilateral polycystic kidneys. **Origin:** Arjumand, Department of Radiodiagnosis, King George's Medical University, Lucknow, India

Figure 5

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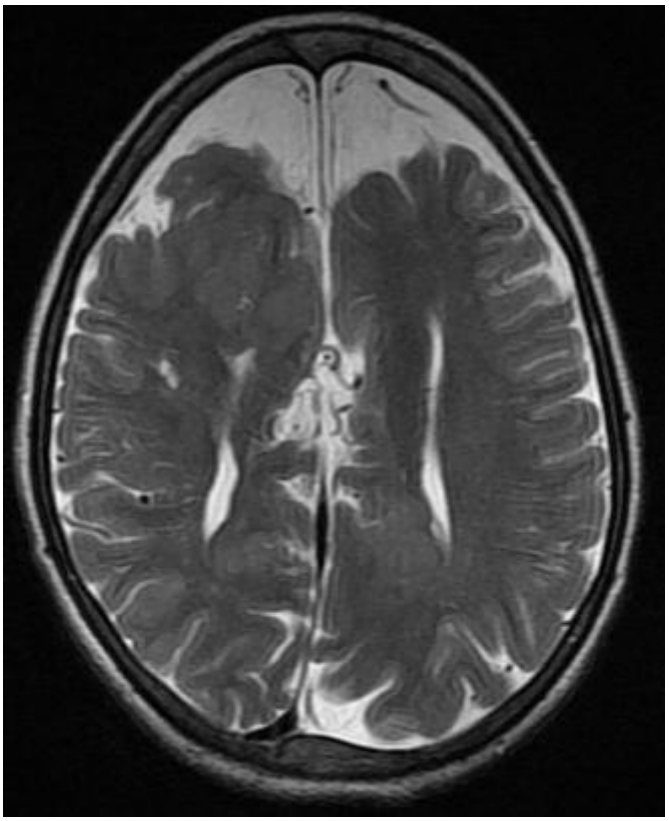

Description: T2 FRFSE axial section of brain showing bilateral frontal lobe atrophy, grey matter heterotopia in right frontal lobe, polymicrogyria in bilateral frontal & parietal lobe with parallel and wide apart body of lateral ventricles. **Origin:** Arjumand, Department of Radiodiagnosis, King George's Medical University, Lucknow, India

Figure 6

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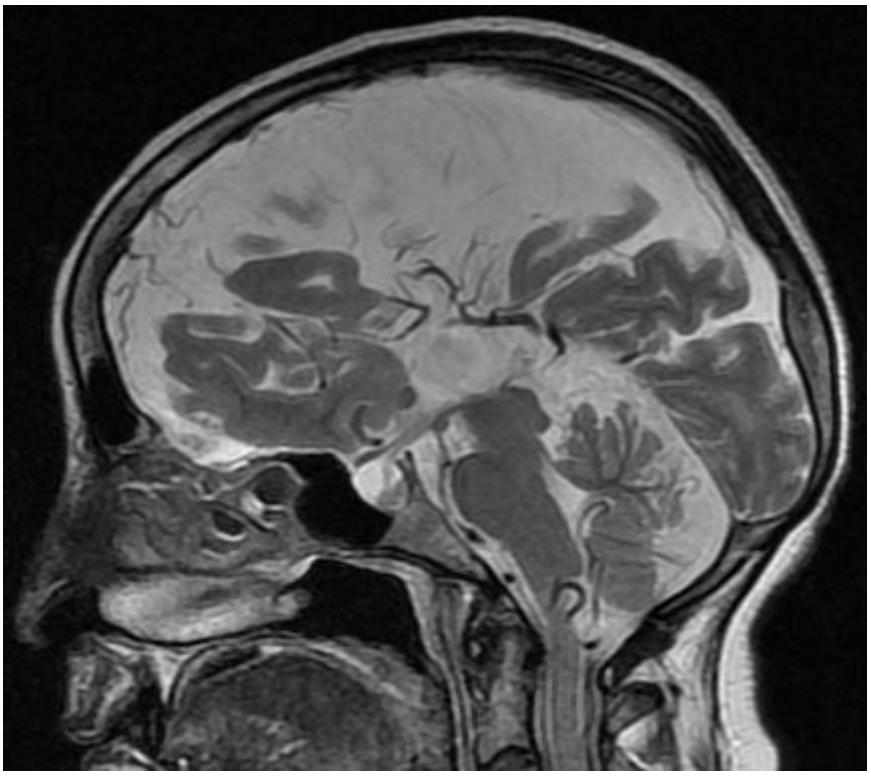

Description: T2 FRFSE sagittal section of brain showing complete agenesis of corpus callosum and age inappropriate atrophy of brain. **Origin:** Arjumand, Department of Radiodiagnosis ,King George's Medical University, Lucknow, India

Figure 7

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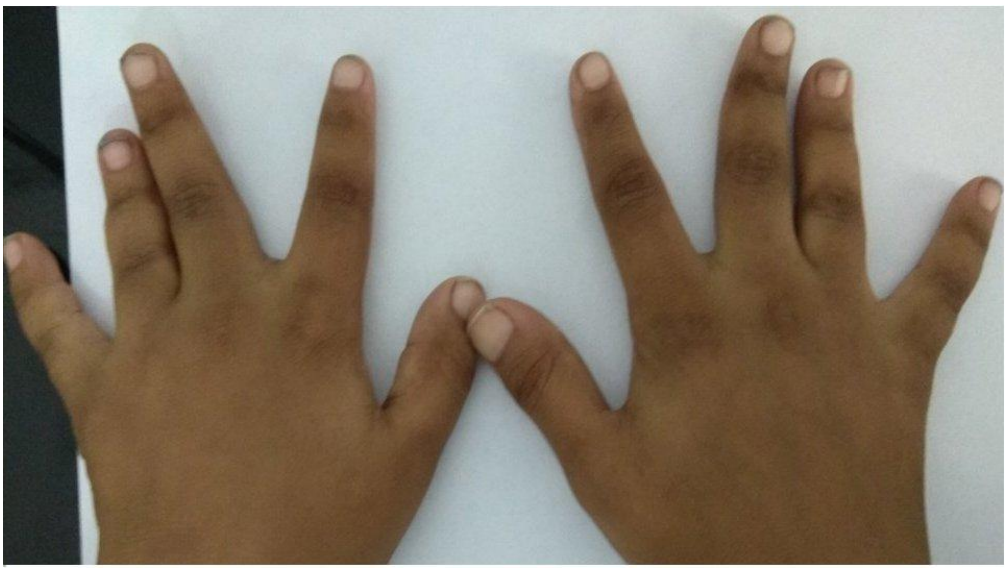

Description: Brachydactyly **Origin:** Arjumand, Department of Radiodiagnosis, King George's Medical University, Lucknow, India

Figure 8

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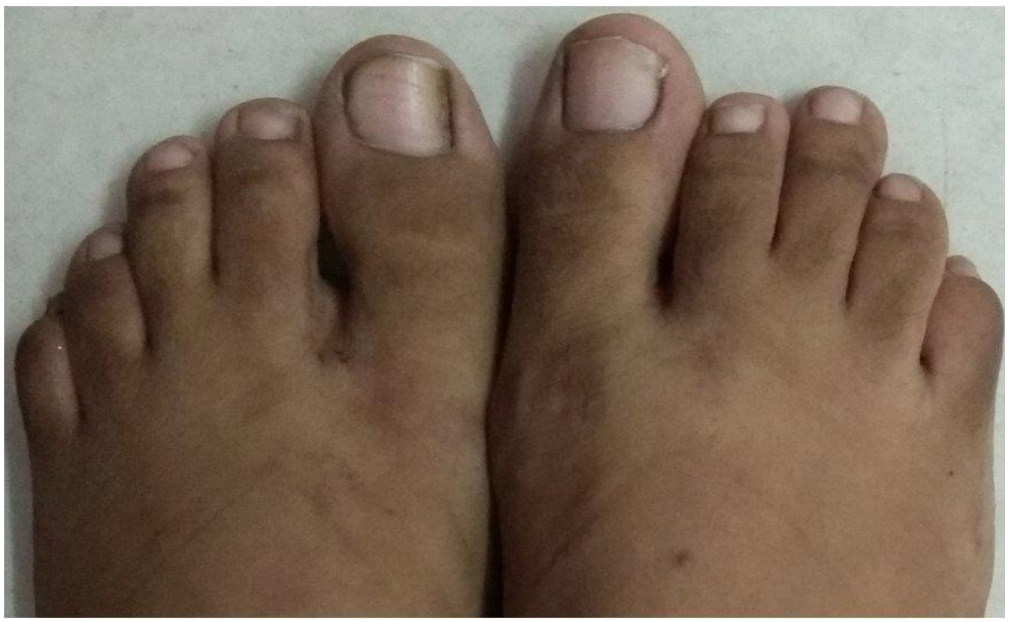

Description: Brachydactyly **Origin:** Arjumand, Department of Radiodiagnosis, King George's Medical University, Lucknow, India