The Thirteenth Case of a Congenital Portosystemic Shunt in a Down Syndrome Patient with Case Review

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INTRODUCTION

- Down syndrome (DS) is caused by non-disjunction, translocation or mosaicism at chromosome 21.
- DS is characterized by intellectual disability, dysmorphic facial features, and congenital malformations including gastrointestinal malformations.
- **Congenital portosystemic shunts** (CPSS) are rare vascular malformations that form **aberrant connections between a portal and systemic vein**.
- **Angiogenic factors** may play a role in the **development of CPSS** as they already contribute to features seen in Down syndrome including placental hypo-vascularity, increased fetal nuchal fold thickness, and tendency to develop pulmonary hypertension.

CASE PRESENTATION

HISTORY:

- Three-year old male presented with hyperammonemia, altered mental status, and choreiform movements of upper and lower extremities.
- History leading up to presentation was largely negative with no sick contacts, no travel history, no recent surgery. Review of Systems was non-contributory.

PMH:

- Atrial septal defect, Gastroesophageal reflux disease status post Nissen fundoplication, Celiac's disease and Failure to thrive.

PHYSICAL EXAM:

- He appeared lethargic. Facial anomalies were consistent with DS. A grade 2/6 systolic murmur was heard on auscultation. Anisocoria was present, with left pupil measuring between 6-7 mm compared to the right pupil at 4-5 mm. Hypotonia of the extremities and trunk were observed.

LABS:

- A comprehensive metabolic panel (CMP) showed a bicarbonate level of 15 mmol/L (ref range 23-32), anion gap of 22 mmol/L (ref range 9-18), aspartate aminotransferase (AST) of 129 U/L (ref range 8-60), alanine aminotransferase (ALT) of 62 U/L (ref rage 6-45) and lactic acid of 10.9 mmol/L (0.7-2.5).
- Blood cultures and Strep-A throat culture yielded no growth. Saliva toxicology screen, salicylate and ethanol levels were normal.
- His **ammonia level was elevated at 231** mol/L (ref range 12-48) and persisted despite the administration of sodium phenylacetate and sodium benzoate.
- Evaluation for metabolic causes were unrevealing.

IMAGING:

- An abdominal ultrasound revealed an **anomalous course of portal venous flow**.
- A CT angiography of the abdomen and pelvis demonstrated a connection between the right portal vein and inferior vena cava, consistent with an intrahepatic congenital portocaval fistula.
- The lower lungs showed dilated pulmonary artery branches of the peripheral system, suggesting developing hepatopulmonary syndrome.

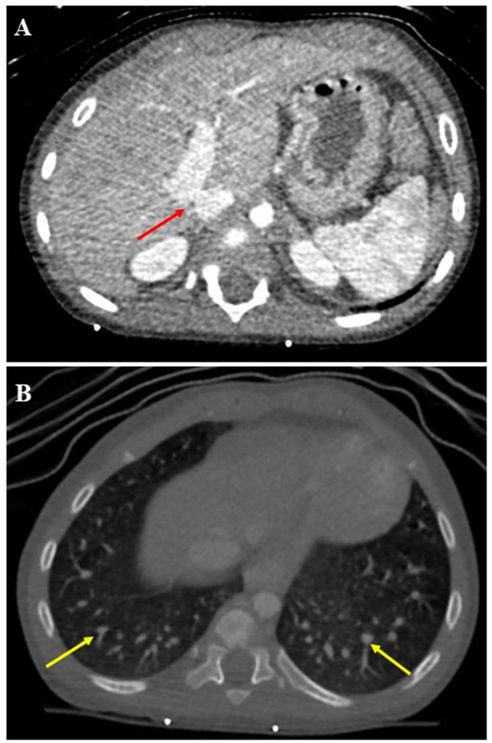

Figure 1: A) A CT of the abdomen and pelvis shows a connection between the right portal vein and inferior vena cava, consistent with CPSS (red arrow). B) Dilated pulmonary artery branches of the peripheral system are seen, suggestive of HPS (yellow arrow).

First Author Year	Chromosomal Abnormality	Age at Diagnosis	Umbilico-Portal venous System Defect	Portosystemic Shunt	Cardiovascular Malformations	Hyperammonemia	Hyperbilirubinemia
Kieran 1992	47, XY, 2/21 translocation	Newborn	Absence of right intrahepatic PV	Right PV to IVC	complete A-V canal defect with PDA	N	Y
Kitagawa 1992	47, XY, +21	Three year old	-	Portal sinus to IVC branch of the proper hepatic artery	AV canal defect and heart block	Y	Y
Mahony 1992	Trisomy 21	Fetus	Varix of UV	-	-	-	-
Estroff 1992	Trisomy 21	Fetus	Varix of UV	-	-	-	-
Challis 1997	Trisomy 21	Fetus	Varix of UV	-	-	-	-
Hartung 2000	47, XY, +21	In utero (14 weeks)	AV fistula between the umbilical vein and hepatic arteries	Patent Ductus Venosus	AV canal defect	-	-
Hartung 2000	47, XY, +21	In utero (31 weeks)	AV fistula between the umbilical vein and hepatic arteries	Patent Ductus Venosus	-	-	-
Courtens 2000	47, XY, +21	Newborn	Absence of ductus venosus	Portal vein to IVC	VSD	N	-
Contratti 2001	Trisomy 21	Fetus	Absence of ductus venosus	-	-	-	-
Pipitone 2003	47, XY, +21	Fetus	Absence of the intrahepatic portal vein and ductus venosus	Portal Sinus to IVC	VSD	Y	Y
Pipitone 2003	47, XY, +21	Newborn	Absence of the intrahepatic portal vein and ductus venosus	Portal Sinus to IVC	VSD and ASD	Y	N
Tatekawa 2005	Trisomy 21	6-years	-	SMA to SMV - AVM	ASD	-	-
Franchi-Abella 2010	Trisomy 21	35-weeks	-	L PV and IVC, intrahepatic	-	N	N
Golewale 2010	Trisomy 21	4-months	Complex Nidus-type shunt	the portal vein that, in turn, drained via a PDV into IVC	ASD, large VSD, large PDA, PDV	-	-
Golewale 2010	Trisomy 21	Newborn	Shunt into a phrenic vein	Patent Ductus Venosus to IVC	PFO, PDA, RVH	-	-
Golewale 2010	Trisomy 21	Newborn	Intrahepatic shunt L PV to L hepatic vein	Patent Ductus Venosus to IVC	ASD, VSD, RVH	-	-
Sokollik 2013	Trisomy 21	Newborn	Type 1	Portal vein to IVC	N	-	-
Sokollik 2013	Trisomy 21	Five month old	Type 2	Portal vein to IVC	AVSD	Y	N
Doan 2014	Trisomy 21	1-month old	Type 2 intrahepatic	L portal v to L hepatic v	N	-	Y
Timpanaro 2015	Trisomy 21	2-year old	Side-to-Side shunt	Portal vein to IVC	PDA	-	-
Losa 2015	Trisomy 21	22-days old	middle hepatic vein and intra- hepatic portal vein	Left portal and left hepatic vein	-	-	Y
Yamaguchi 2016	47, XX, +21	Newborn	Patent ductus venosus anomalous connection	Portal vein to IVC	N	Y	Y
Nohomovich 2022	Trisomy 21	Three year old	Type 1	Right PV to IVC	Y	Y	Y

Table 1: Reported cases with down syndrome and congenital portosystemic shunt. N = No, Y = Yes, - = not reported.

OUTCOMES

- Interventional radiology placed an 18 mm, Amplatzer PFO closure device within the congenital fistula.
- There was no residual flow seen in the portal vein from the inferior vena cava post-procedure.
- Clinical improvement was noted following the procedure. His ammonia levels normalized. His mental status returned to baseline with resolution of choreiform movements. A follow-up abdominal ultrasound demonstrated no residual or recurrent portocaval fistula.
- At two-years follow-up, the patient had no recurring issues related to the fistula or closure device placement.

CLINICAL PEARLS

- Consider CPSS in DS patient that present acutely with hyperammonemia.
- Patient's might have hyperbilirubinemia acute hepatitis depending on shunt location and presentation.
- Patient's with CPSS will often have cardiac abnormalities, consider an echocardiogram.
- Imaging is important for the diagnosis of CPSS and an ultrasound with doppler can be performed but a CT should be done as well to further assess the vasculature.
- Surgical correction if often curative for the hyperbilirubinemia.

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