cholesterolosis is rare. Gall bladder metaplasia, dysplasia and incidental GBC are infrequently seen in South India.

CONFLICTS OF INTEREST

The authors have none to declare.

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CLINICAL PROFILE OF PATIENTS WITH SIMULTANEOUS PRESENTATION OF WILSON'S DISEASE AND AUTOIMMUNE LIVER DISEASE IN A RURAL TERTIARY CARE CENTRE

R. Ramasubramanian [1,*], Y. Jaipaul [1], A. Shafique [1], Poppy Rejoice [2]

[1] *Thoothukudi Medical College, Thoothukudi, India*
[2] *KanyaKumari Government Medical College, Asaripallam, India*
E-mail address: ramlalli90@yahoo.com
(R. Ramasubramanian).

Background and Aim: The coexistence of Wilson's disease and autoimmune liver disease in a same patients is a rare entity. In this situation, combined treatment with steroid and D-Penicillamine may be effective.

Methods: We analysed clinical, histological, laboratory profile for patients with chronic liver disease with aim of finding the etiology of the disease after ruling out common causes like alcohol, viruses and drugs.

Results: Out of 10 patients 6 were males and 4 were females. Commonest clinical presentation was abdominal distension (80%), abdominal pain (30%), pedal edema (60%), splenomegaly (40%) and upper GI bleed (40%). Laboratory investigation revealed anemia (50%), thrombocytopenia (70%), prothrombin time prolongation in (60%), normal liver function in 60%, abnormal liver function in (40%). Hepatitis A, B, C and E were negative in all the cases. Serum cereloplasmin <20 mg% in (30%), normal level in (70%). 24 h urinary copper level range of >200 in (40%), 100 to 200 in (40%) and 90 to 100 in (20%). Autoimmune markers revealed ANA strong positivity in (40%), mild positivity in (60%). AMA, ASMA, Anti-LKM-1 were negative in all cases (100%). Liver biopsy showed features of Autoimmune liver disease and PERIPORTAL copper deposition in 80% of cases where 20% showed features of copper deposition in liver.

Conclusion: The coexistence of Wilson's disease and autoimmune liver disease is a rare entity and clinician should have high level of suspicion in diagnosing the problem and medical treatment with steroids and D-Pencillamine simultaneously to be started in these patients (Figure 1).

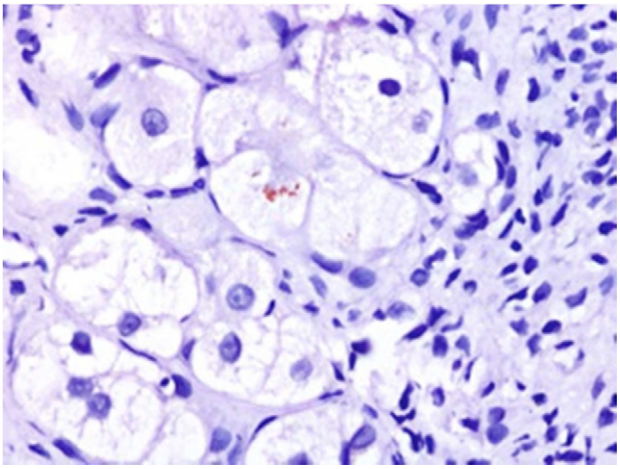

Figure 1. Liver biopsy.

CONFLICTS OF INTEREST

The authors have none to declare.

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MOLECULAR DIAGNOSIS OF WILSON'S DISEASE USING NEXT GENERATION SEQUENCING PLATFORM

Rekha Athiyarath, Aaron Chapla, Madeline Vidhya, Ashish Goel*, C.E. Eapen, Uday Zachariah, Nihal Thomas, Thomas Paul, Nithin Kapoor

Christian Medical College, Vellore, India
E-mail address: drashishgoel@cmcvellore.ac.in (A. Goel).

Background and Aim: In patients with hepatic Wilson's disease (WD), confirmatory diagnosis is often elusive. Currently available genetic tools for WD diagnosis are time consuming and expensive. Next generation sequencing (NGS) offers high throughput sample processing at a relatively lower cost. Therefore, we aim to investigate the feasibility of the NGS-based approach to sequence *ATP7B* gene in patients with hepatic WD.

Method: Patients diagnosed with hepatic WD (low serum Ceruloplasmin, high urinary copper excretion and presence of Kayser-Fleischer ring on eye examination) were included after informed consent and blood was collected for DNA extraction. All 21 exons and flanking 5′ and 3′ UTR regions of *ATP7B* gene were amplified in four multiplex PCRs. Libraries were generated from the pooled amplicons and subjected to sequencing on Ion-Torrent platform. Data analysis was performed using IonTorrent Suite™ Software v.5.0 and genetic variants were then identified using Torrent Suite Variant Caller pipeline v.5.0. Bioinformatic tools (SIFT, Polyphen, Muta-

Table 1 Demographic Characteristics and Mutation Details in Patients With Wilson's Disease.

UPN No	Age	Sex	Location	ATP7B mutation	Aminoacid change	Exon	SIFT	Polyphen	MT
WD1	16	M	JH	c[3895C>T] c.3182G>A]	p.L1299F[a] G1061E	18,4	Damaging	Probably damaging	Disease causing
WD2	24	F	TN	c.[813C>A]	p.C271x	2	–	–	Disease-causing
WD3	26	F	TN	c.[3971A>G]	p.N1324S	19	Tolerated	Probably damaging	Disease-causing
WD4	24	F	TN	c[3971A>G]	p.N1324S	19	Tolerated	Probably damaging	Disease-causing
WD5	23	M	WB	c[3182G>A] c[2131G>T]	p.G1061E G711W	14,8	Damaging	Probably damaging	Disease-causing
WD6	16	F	AP	c[2131G>T]	p.G711W	8	Damaging	Probably damaging	Disease-causing
WD7	9	M	WB	c[3662G>T] c[3053C>T]	p.G1221V[a] p.A1018V	17,13	Tolerated Damaging	Probably damaging	Disease-causing
WD8	36	M	BGD	NEGATIVE	–	–	–	–	–
WD9	27	M	WB	NEGATIVE	–	–	–	–	–
WD10	17	M	WB	c.[3567delT]	p.C1189fs[a]	17	–	–	Disease-causing
WD11	15	M	UP	C[813C>A[=]	p.C271X[=]	2	–	–	Disease-causing
WD12	22	M	WB	c.[3182G>A]	p.G1061E	14	Damaging	Probably damaging	Disease-causing
WD13	17	M	WB	c.[3802G>A] c.[813C>A]	p.G1268R, p.C271x	18,2	Damaging	Probably damaging	Disease-causing

Geographic Location: JH-Jharkhand, WB-West Bengal, TN-Tamilnadu, UP-Uttar Pradesh, AP-Andhra Pradesh, BGD-Bangladesh.
[a] Novel mutation.

tion taster) were used to predict the effect of the observed variants. Sanger sequencing was subsequently performed in the corresponding exon to confirm the identified mutations.

Results: Thirteen patients with unequivocal hepatic WD were included for the study (Table 1). Coverage analysis showed >99% of the target was sequenced with a mean coverage of 20x. Overall we identified 9 mutations (missense: 7 nonsense: 1, deletion: 1) in 11 patients (84.6%). Four patients were compound heterozygous, six were homozygous and one was heterozygous for mutations (Table 1). Among the identified mutations, three were novel and all others have been previously reported. We failed to identify point mutations in two patients. Sanger sequencing results were 100% concordant with NGS data.

Conclusion: In this pilot study, we report the use of NGS platform for *ATP7B* gene mutation analysis for molecular diagnosis of WD. Our data suggests NGS is a reliable method for the diagnosis of Wilson's disease.

CONFLICTS OF INTEREST

The authors have none to declare.

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SPONTANEOUS RESOLUTION OF POST-TRAUMATIC HEPATIC ARTERY PSEUDOANEURYSM PRESENTING AS HEMOBILIA, HEMOCHOLECYSTITIS AND SHOCK

Sajan Agarwal*, Madhur Jain, Raj K. Gupta

SMS Medical College, Jaipur, India
E-mail address: drsajanagarwal@yahoo.com (S. Agarwal).

Background: Post-traumatic hepatic artery pseudoaneurysm is a rare, but life threatening condition. Symptoms can occur days or even months after the trauma.

Case Summary: A 12-year-old child was presented to us with complaints of hematemesis, melena and severe right hypochondrium colicky pain for last 2 days. He had a history of blunt abdominal trauma (accidental fall from height) 1-month back requiring admission in outside hospital. At the time of admission to us, the child was hemodynamically unstable. CECT abdomen showed a focal area (14 × 8 mm) of staining in arterial phase was seen in segment 5, suggestive of pseudoaneurysm of right hepatic artery branch. CT angiography confirmed the presence of pseudoaneurysm. He was resuscitated and after hemodynamic stabilization, our interventional cardiology team planned angiographic embolization of pseudoaneurysm. On 3rd day of admission, child developed chicken pox and procedure was deferred for sometime. Repeat CECT Abdomen with CT Angiography (done 10 days after admission and approx 6 wks after trauma) showed complete resolution of pseudoaneurysm. Child