Wilson disease: Canadian perspectives on presentation and outcomes from an adult ambulatory setting

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SUPPLEMENTARY TABLES

SUPPLEMENTARY TABLE 1
Leipzig criteria used to establish appropriateness of Wilson disease diagnosis (3)

Clinical parameter	Finding	Leipzig score
Kayser-Fleicher rings	Present	2
	Absent	0
Neurological symptoms or classic MRI changes	Severe	2
	Mild	1
	Absent	0
Liver copper concentration	>250 µg/g (>3.93 µmol/g)	2
	50–250 µg/g (0.79–3.93 µmol/g)	1
	<50 µg/g (<0.79 µmol/g)	−1
Coomb's negative hemolytic anemia	Present	1
	Absent	0
Ceruloplasmin	<0.10 g/L	2
	0.1–0.2 g/L	1
	>0.2 g/L	0
Urinary copper	>2 × ULN (>1.2 µmol/day)	2
	1–2 × ULN (0.6–1.2 µmol/day)	1
	Normal (<0.6 µmol/day)	0
	>5 × ULN after 24 h D-penicillamine challenge	2
Mutation analysis	2 identified mutations	4
	1 identified mutation	1
	0 identified mutations	0

ULN Upper limit of normal

SUPPLEMENTARY TABLE 2
Most recently available laboratory values according to presentation type for the cohort

Presentation	Final ceruloplasmin, g/L	Final urinary copper, µmol/24 h	Final serum copper, µmol/L	Final ALT, U/L	Final AST, U/L	Final total bilirubin, µmol/L
Asymptomatic (n=14)	0.06 (UD–0.11)	1.06 (0.47–18.8)	3.35 (0.8–8.0)	37.5 (7–88)	27.5 (15–78)	12.5 (6–22)
Hepatic (n=13)	0.03 (UD–0.15)	3.12 (0.33–6.79)	1.65 (0.6–13.3)	35 (9–204)	33 (15–146)	11 (4–49)
Neurological (n=15)	0.11 (UD–0.18)	1.91 (0.24–15.8)	6.4 (0.2–13)	20 (11–50)	24 (15–38)	9 (5–14)
Hepatic and neurological (n=6)	0.11 (0.04–0.26)	2.55 (1.17–5.71)	4.8 (1.2–16.7)	30 (16–100)	35 (16–115)	11 (7–61)
Total (n=48)	0.05 (UD–0.26)	1.95 (0.25–18.8)	3.35 (0.6–16.7)	30 (7–204)	30 (15–146)	10 (4–61)

Data presented as median (range). At time of most recent set of values, median time on treatment was 10.1 years (range 0 to 43.7 years). ALT Alanine aminotransferase; AST Aspartate aminotransferase; UD Undetectable

SUPPLEMENTARY TABLE 3
Ultrasound results according to presentation type of 42 patients who underwent abdominal ultrasound imaging

	Ultrasound		Coarse echotexture		Cirrhosis		Splenomegaly (or other evidence of portal hypertension)	
Presentation	D	FU	D	FU	D	FU	D	FU
Asymptomatic (n=14)	2	12	1	2	0	2	1	2
Hepatic (n=13)	3	12	0	3	3	6	2	8
Neurological (n=15)	5	8	2	6	2	0*	4	4
Hepatic and neurological (n=6)	3	5	1	1	1	2†	1	3
Total (n=48)	13	37	4	12	6	10	8	17

*Data presented as n. Results are divided into patients with diagnostic ultrasounds before and up to one-year post-diagnosis (D) and those with follow-up (FU) ultrasounds performed one year or more postdiagnosis (some patients had both D and FU ultrasounds performed). Median time between D ultrasounds and treatment initiation, and most recent FU ultrasound and treatment initiation was 12 days (range −54.8 months to +12.0 months) and 13.3 years (range 1.5 to 41.8 years), respectively. Course echotexture was only counted if it was documented in the ultrasound report, but there was no documentation of actual cirrhosis. Cirrhosis was counted if it was specifically mentioned in the report and includes mild-severe levels. Splenomegaly was defined as a spleen span >12.0 cm and other evidence of portal hypertension including documented splenic varices or reversal of portal venous flow. *Four patients had cirrhosis reported on a previous ultrasound, but not their most recent; †The cirrhosis seen for one of these patients refers to his last pretransplant ultrasound*

SUPPLEMENTARY TABLE 4
Upper gastrointestinal endoscopy results according to presentation type of 22 patients who underwent endoscopic imaging

	Endoscopy		Portal hypertensive gastropathy		Esophageal varices		Gastric varices	
Presentation	**D**	**FU**	**D**	**FU**	**D**	**FU**	**D**	**FU**
Asymptomatic (n=14)	2	3	1	3	2	2	0	1
Hepatic (n=13)	3	5	3	1	1	0	1	0
Neurological (n=15)	1	5	0	0	0	1	0	0
Hepatic and neurological (n=6)	2	3	1	0	0	0	0	0
Total (n=48)	8	16	5	4	3	3	1	1

Data presented as n. Results are divided into patients with diagnostic (D) endoscopies, before and up to one year postdiagnosis, and those with follow-up (FU) endoscopies one year or more postdiagnosis (some patients had both D and FU endoscopies performed). Median time between D endoscopies and treatment initiation, and most recent FU endoscopies and treatment initiation was −15 days (range −3.8 to 1.1 months) and 9.8 years (range 1.5 to 41.6 years), respectively. Portal hypertensive gastropathy was counted if it was documented in the imaging report and refers to any grade of gastropathy