Genetic Diagnostic and Research Laboratory Department of Anatomy Faculty of Medical Sciences University of Sri Jayewardenepura Nugegoda.

CONFIDENTIAL MOLECULAR GENETIC LABORATORY REPORT.

Name of the patient	Age/sex	null/null
Date of collection	Date of reporting,	12/13/15 7:05 PM
Case ref no	Referred By	
add Referred By : HD		

REPORT ON HUNTINGTON'S DISEASE

Methodology:10 ml blood was collected from the patient and DNA was extracted from the buffy coat of the EDTA anticoagulated blood. The mutation was characterized by PCR using primers flanking the CAG repeat region and analysed on Gene Scan software.

Results and counseling: On analysis the patient showed

	Triplet Repeat Size		
	Allele 1	Allele 2	Normal range
HD	166.66667	14248.332	11-34

Comments:

1. Huntington disease is caused due to expansion of triplet repeats in HD gene.

2. Reference CAG repeat values are,

Normal Allele: ≤ 26 CAG repeats Intermediate Allele: 27-35 CAG repeats

- 3. This test detects expansion of triplet repeats in HD gene. Thus a positive test report confirms Huntington disease
- 4. Advised Genetic Counseling