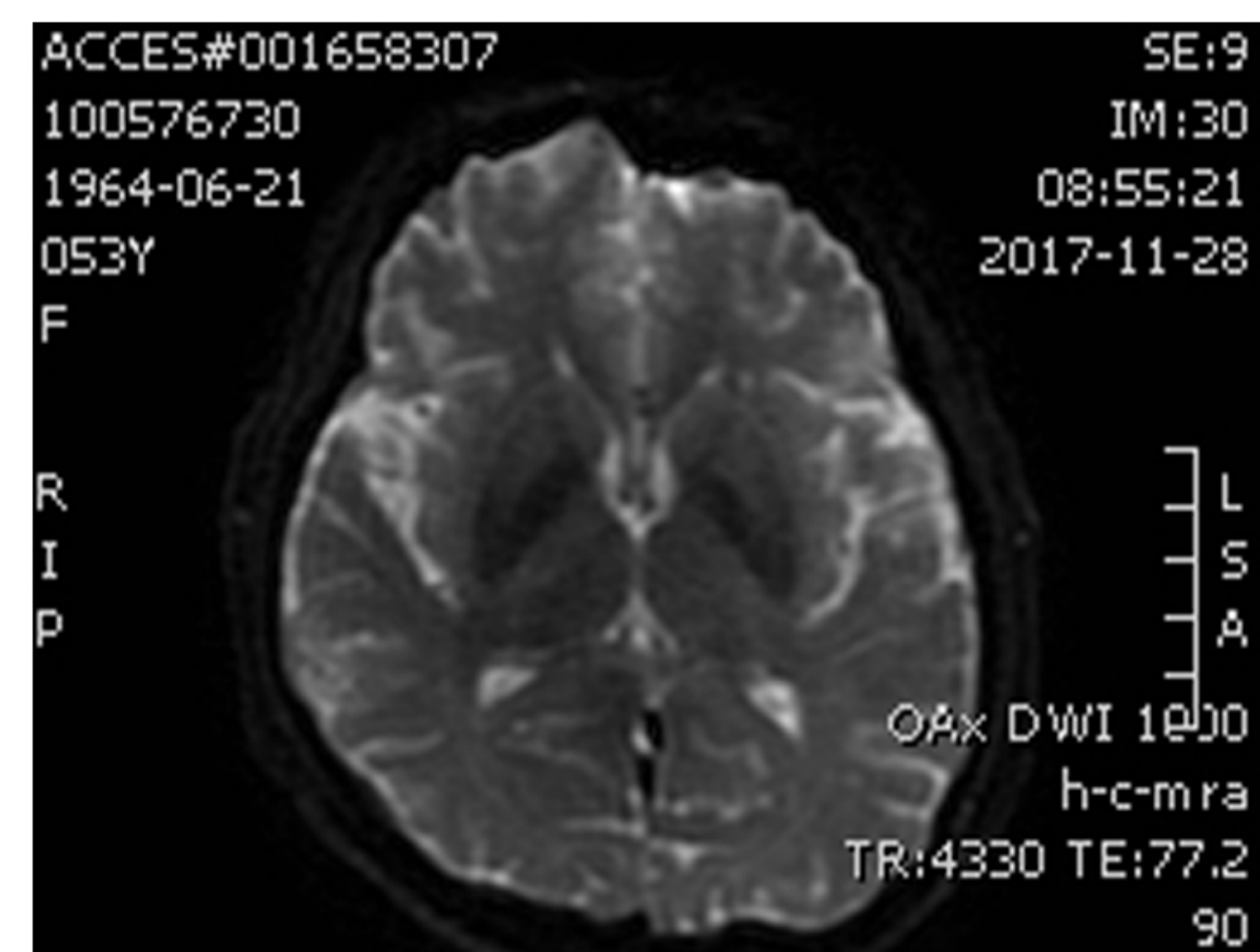
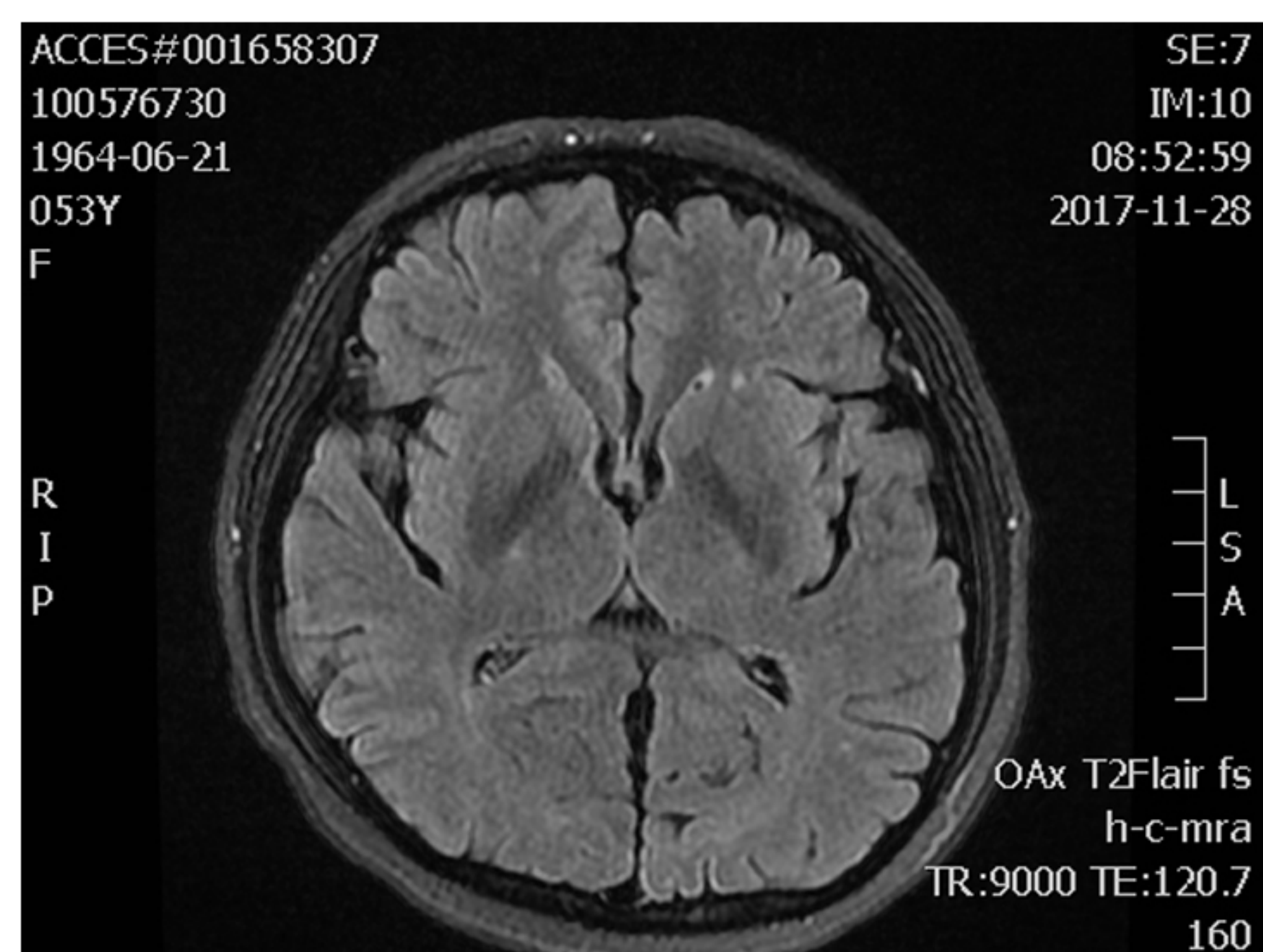
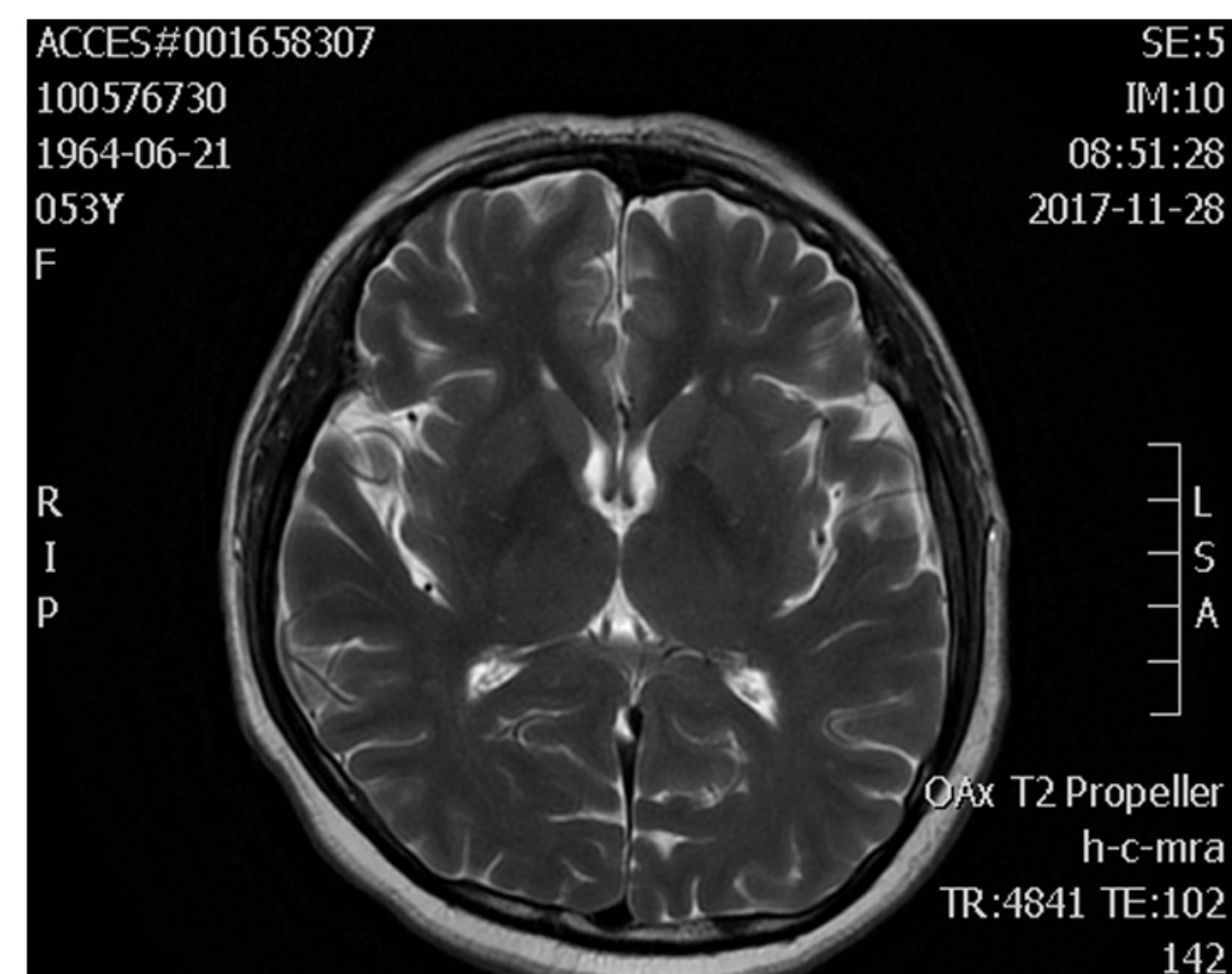
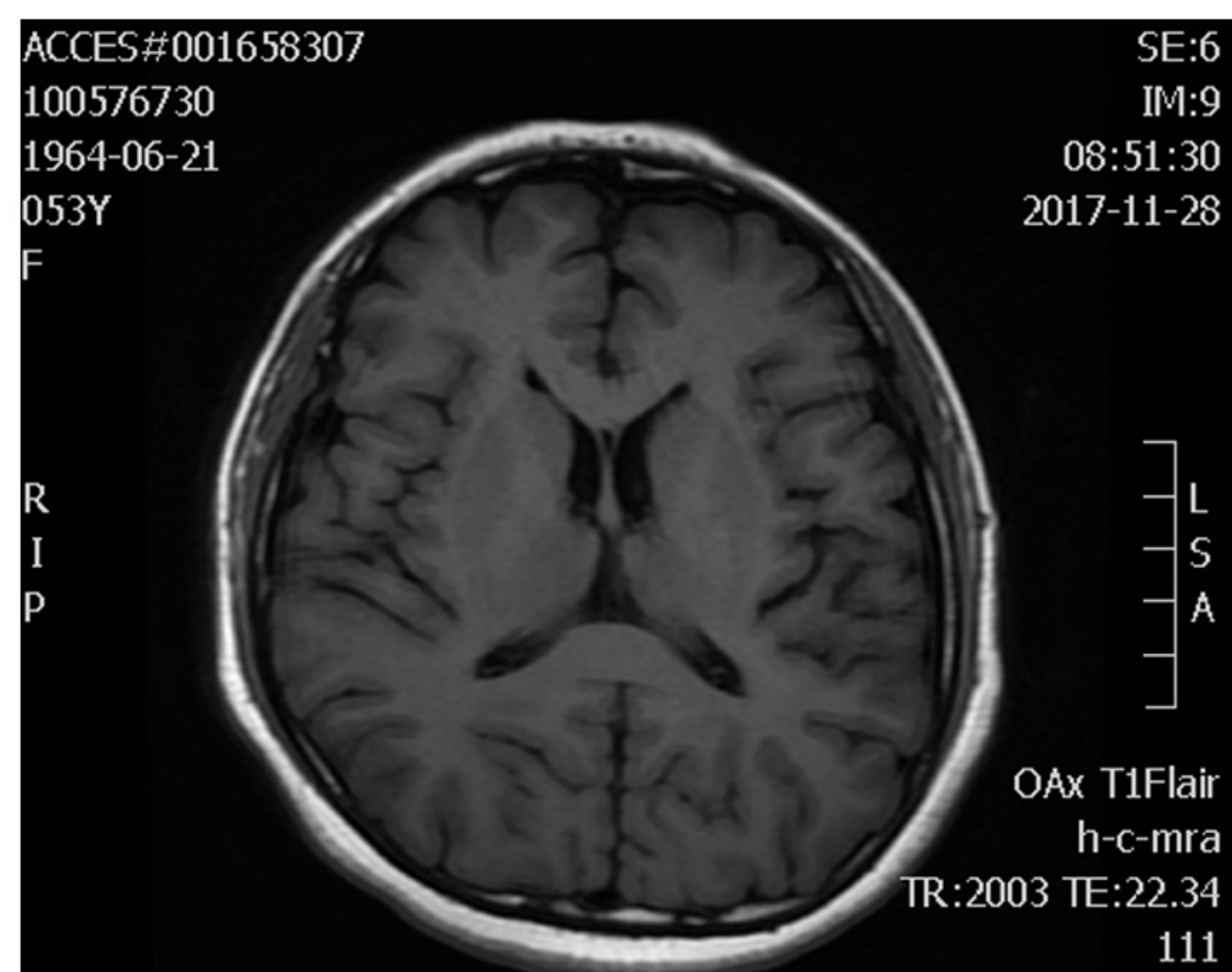
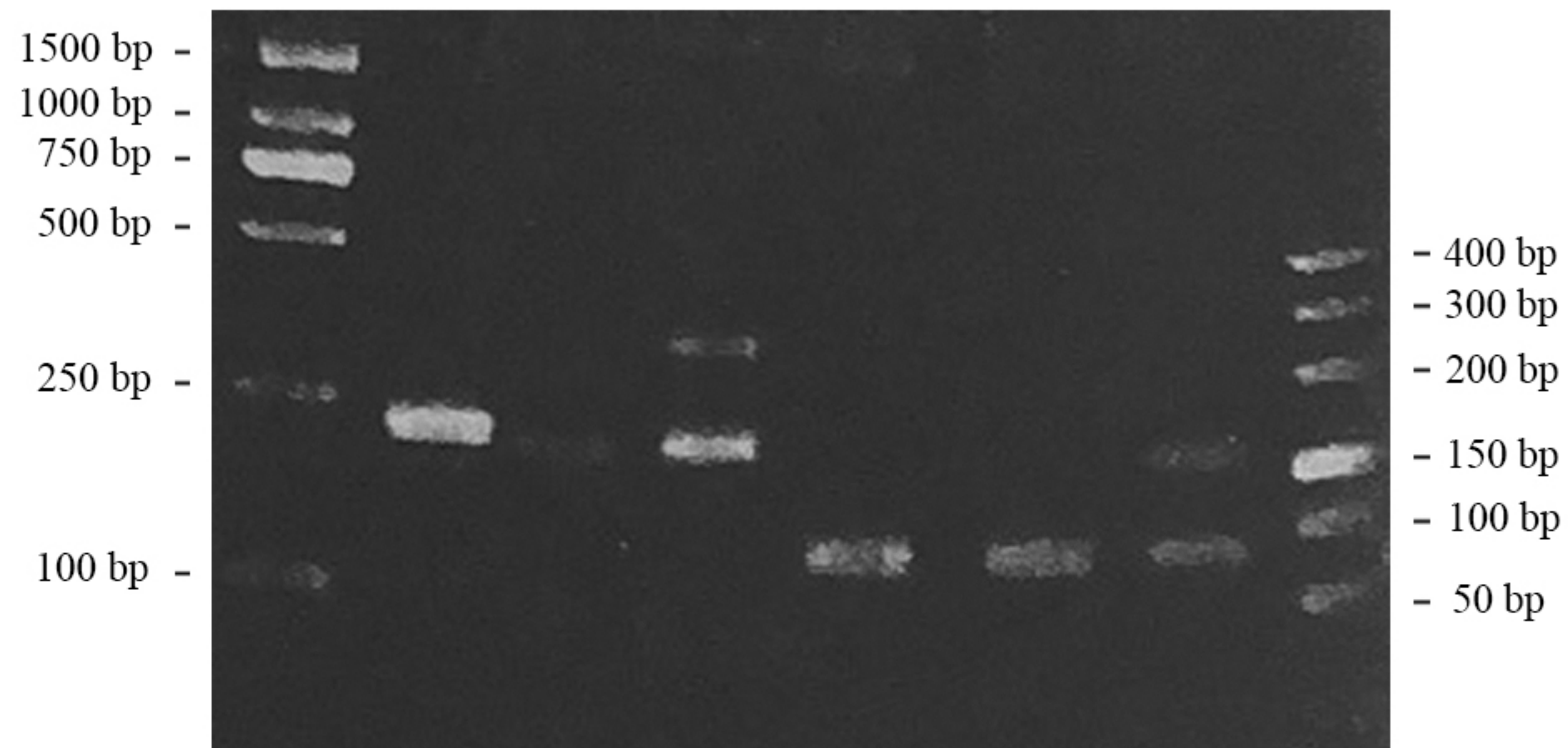






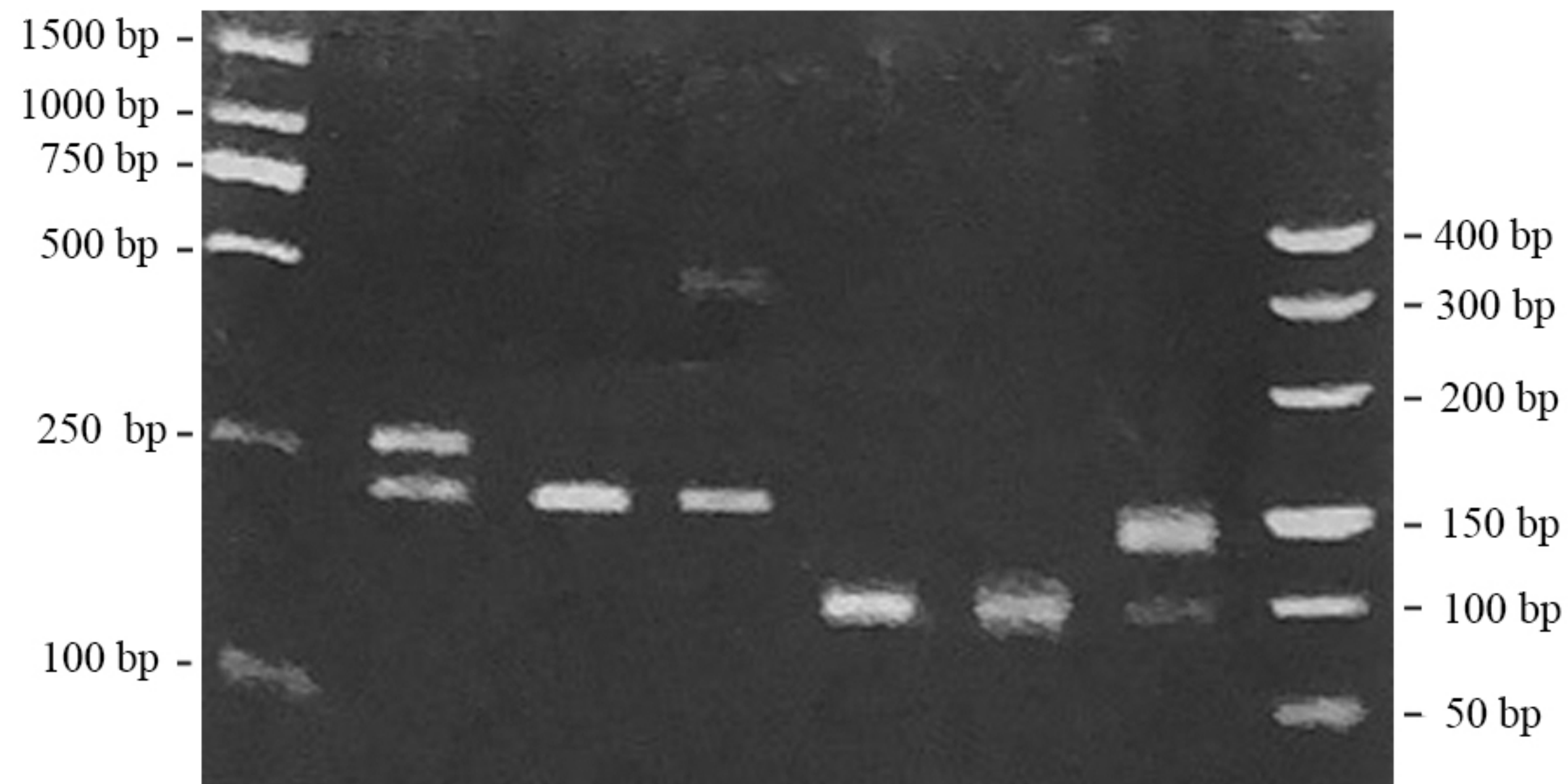
Negative control  
 Proband  
 Positive control  
 Negative control  
 Proband  
 Positive control



ATXN1 EX8

ATXN2 EX1

Negative control  
 Proband  
 Positive control  
 Negative control  
 Proband  
 Positive control



ATXN3 EX12

CACNA1A EX48

Table S1 Needle EMG results of the proband

| Side  | Muscle                            | Insertional | Spontaneous Activity |        | MUAP   |          |          | Poly | Recruitment  |
|-------|-----------------------------------|-------------|----------------------|--------|--------|----------|----------|------|--------------|
|       |                                   | Activity    | Fib                  | PSW    | Fas    | Dur      | Amp      |      |              |
| Right | Paraspinal muscle C8              | Increase    | 1+                   | Normal | Normal | Increase | Increase | None | Reduced      |
| Left  | Rectus abdominis                  | Increase    | Normal               | 2+     | Normal | Increase | Increase | None | Reduced      |
| Left  | Sternocleidomastoid               | Normal      | Normal               | Normal | Normal | Normal   | Normal   | None | Interference |
| Left  | Extensor indicis                  | Increase    | Normal               | 1+     | Normal | Increase | Increase | None | Reduced      |
| Left  | Biceps                            | Increase    | 1+                   | 2+     | Normal | Increase | Increase | None | Reduced      |
| Right | Extensor digitorum                | Increase    | 2+                   | 1+     | 1+     | Increase | Increase | None | Reduced      |
| Right | Short abductor of pollicis brevis | Increase    | 3+                   | 1+     | Normal | Increase | Increase | None | Reduced      |
| Right | Dorsal interossei                 | Increase    | 2+                   | Normal | Normal | Normal   | Normal   | None | Reduced      |
| Right | Vastus medialis                   | Normal      | -                    | Normal | Normal | Normal   | Normal   | None | Interference |
| Right | Tibialis anterior                 | Increase    | Normal               | 1+     | Normal | Normal   | Normal   | None | Reduced      |
| Left  | Gastrocnemius                     | Normal      | Normal               | Normal | Normal | Normal   | Normal   | None | Reduced      |

Fib, Fibrillation; PSW, Positive sharp waves; Fas, Fasciculation; Dur, Duration; Amp, Amplitude; Poly, Polyphasic.

Table S2. Nerve conduction studies summary of the proband

| Side                                                       | Nerve       | Latency (ms) | Amplitude (mv) | Conduction velocity (m/s) |
|------------------------------------------------------------|-------------|--------------|----------------|---------------------------|
| Tibial Motor                                               |             |              |                |                           |
| Left                                                       | Wrist-ADM   | 2.38         | 3.6            | NA                        |
|                                                            | Elbow-Wrist | 6.63         | 2.8            | 52.9                      |
| Tibial Motor                                               |             |              |                |                           |
| Right                                                      | Wrist-ADM   | 2.54         | 3.7            | NA                        |
|                                                            | Elbow-Wrist | 6.56         | 3.5            | 59.7                      |
| Median Motor                                               |             |              |                |                           |
| Left                                                       | Wrist-APB   | 4.15         | 1.35           | NA                        |
|                                                            | Elbow-Wrist | 8.71         | 1.14           | 51.6                      |
| Median Motor                                               |             |              |                |                           |
| Right                                                      | Wrist-APB   | 3.85         | 2.2            | NA                        |
|                                                            | Elbow-Wrist | 7.60         | 1.89           | 53.3                      |
| ADM, Abductor digiti minimi; APB, Abductor pollicis brevis |             |              |                |                           |

Table S3. Motor neuron disease (MND)-related genes

| Gene     | NM number    | Gene    | NM number    |
|----------|--------------|---------|--------------|
| ABCA2    | NM_212533    | MYBPC1  | NM_002465    |
| AFG3L2   | NM_006796    | MYH2    | NM_017534    |
| AKR1C3   | NM_003739    | MYH7    | NM_000257    |
| ALAD     | NM_000031    | NDUFV1  | NM_007103    |
| ALDH18A1 | NM_002860    | NEFH    | NM_021076    |
| ALS2     | NM_020919    | OPA3    | NM_025136    |
| AMPD2    | NM_004037    | OPTN    | NM_001008213 |
| ANAPC7   | NM_016238    | PARK7   | NM_001123377 |
| ANG      | NM_001145    | PFN1    | NM_005022    |
| ANO10    | NM_018075    | PLP1    | NM_001128834 |
| AP4E1    | NM_007347    | PNPLA6  | NM_001166111 |
| ATXN2    | NM_002973    | PRPH    | NM_006262    |
| BEAN1    | NM_001136106 | PRPH2   | NM_000322    |
| BSCL2    | NM_001122955 | RANBP2  | NM_006267    |
| c19orf12 | NM_031448    | SCN4A   | NM_000334    |
| C9orf72  | NM_018325    | SCN9A   | NM_002977    |
| CACNA1A  | NM_023035    | SETX    | NM_015046    |
| CHCHD10  | NM_213720    | SLC52A3 | NM_033409    |
| CHMP2B   | NM_014043    | SMN1    | NM_000344    |
| CYP7B1   | NM_004820    | SMN2    | NM_022875    |
| DAO      | NM_001917    | SOD1    | NM_000454    |
| DCAF8    | NM_015726    | SPAST   | NM_014946    |
| DCTN1    | NM_004082    | SPG11   | NM_025137    |
| DSCAML1  | NM_020693    | SPG20   | NM_001142296 |
| DYNC1H1  | NM_001376    | SPG21   | NM_016630    |
| EEF2     | NM_001961    | SPG7    | NM_003119    |
| ELOVL4   | NM_022726    | SQSTM1  | NM_003900    |
| ELOVL5   | NM_021814    | SYNE1   | NM_182961    |
| ELP3     | NM_018091    | TAF15   | NM_139215    |
| ERBB4    | NM_005235    | TARDBP  | NM_007375    |
| EXOSC3   | NM_016042    | TBK1    | NM_013254    |
| FIG4     | NM_014845    | TRPM7   | NM_017672    |
| FUS      | NM_004960    | UBQLN2  | NM_013444    |
| GARS     | NM_002047    | UNC13A  | NM_001080421 |
| GRN      | NM_002087    | VAPB    | NM_004738    |
| HDAC6    | NM_006044    | VCP     | NM_007126    |
| HEXB     | NM_000521    | VEGFA   | NM_016516    |
| HNRNPA1  | NM_002136    | VPS54   | NM_016516    |
| HSPB1    | NM_001540    | VRK1    | NM_003384    |
| HSPB3    | NM_006308    | DCAF8   | NM_015726    |
| HSPB8    | NM_014365    | LMNB1   | NM_005573    |
| PLEKHG5  | NM_198681    | SIGMAR1 | NM_005866    |
| MAPT     | NM_001123066 | MATR3   | NM_018834    |

ERLIN2  
TECPR2

NM\_007175  
NM\_014844

GAP43

NM\_001130064

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