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가 ,  
DNA , Werner heli-  
case DNA  
가 가

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50 ~ 60  
8 8)  
1904 , Otto Werner 가 ,  
4  
20) 2,3,9)  
1934 , Oppenheimer Kugel ,  
16), 1000 가 ,  
가  
20 ~ 30

:  
28

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(Cushinoid appearance).

1.

Werner 1917 (2)  
1999 1,250 가  
940 가 가 가 ,  
7) 70%

(hyperkeratosis)

가 가  
Meguro  
15)

2.

1)

Gotto 7) 가 가  
( , ),  
가 ( , , , ,  
, ( , , ), ,  
( , , , , (3)  
, 80% 30 가  
( , , ) 47가 70% 36 5)  
, 15%

2)

가 5)

(4)

(adolescent growth spurt)

(122 ~ 161 cm). 40

80%

T

NK

(1)

4,13)

(pinched nose) 가

anti-dsDNA, ANA,

Rheumatoid factor

(bird-like face) 가 (Fig.1).

4)

Grave's dis-

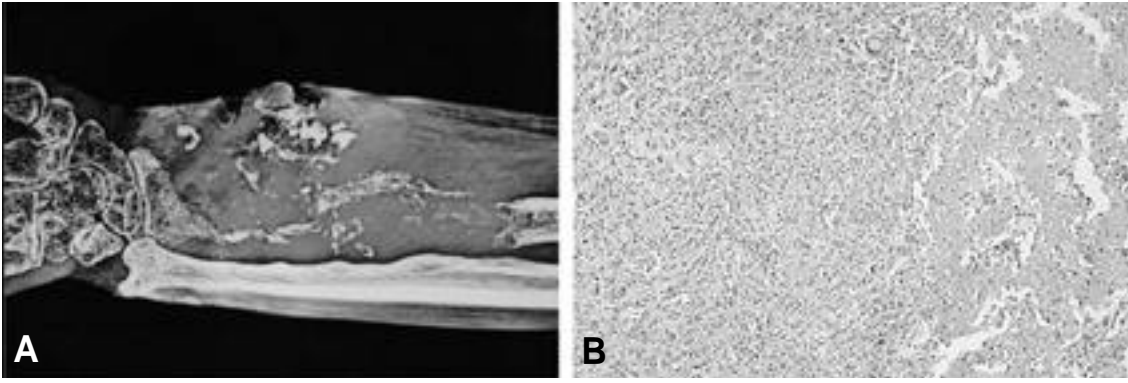
가 ease, SLE 가

가 가 10 ,  
 가 2.3:1 가  
 (5) Papillary, follicular, anaplastic type  
 78%, 14%, 2% ,  
 CT MRI 가 35%, 48%, 13% 6).  
 40 가 6). , (2)  
 40% 가 6). ,  
 10% 37 가 6).  
 가 40 ,  
 가 6).  
 Ishikawa 11) ,  
 10 가 7 , 2 (Fig.  
 (6) 2A,B), 1 Tsuji  
 10) 가 19) 가 55

5), Epstein  
 3 1 가  
 2) 가 가  
 5).  
 Epstein 2)  
 가  
 17).  
 3.  
 1) 가  
 6).  
 (1) 가 6).  
 39



**Fig. 1.** Photograph of the 50 year-old patient, demonstrating characteristic features of Werner syndrome. She looks very old for her ages. She had a cataract on her left eye and a small and beak-shaped nose. (This picture was presented by Yuichi Ishikawa and Makoto Goto.)



**Fig. 2. A.** Radiograph of the forearm of a Werner syndrome showed aggressive bone forming tumor in the left distal radius. (This picture was presented by Yuichi Ishikawa and Makoto Goto.)

**B.** The pathology of this patient was diagnosed as osteosarcoma. It showed hyperchromatic and dysplastic cells surrounded by tumor osteoid matrix. (This picture was presented by Yuichi Ishikawa and Makoto Goto.)

가 (Fig. 2B),

Ishikawa

가

가, (2)

Werner helicase(DNA

<sup>11)</sup> helicase RecQ family)

2)

Werner helicase

DNA

(1)

, DNA

DNA

Boyd Grant <sup>1)</sup>

가

DNA

<sup>6)</sup>

가

가

가

1/3

1/8

4.

(WRNp)

<sup>14)</sup>

, Goto <sup>7)</sup>

가

10

telomere

가 ,  
 chromosome  
 (TTAGG)<sub>n</sub>  
 가 ,  
 가 .  
 telomerase RNA dependent DNA  
 polymerase 가 ,  
 telomerase  
 WRNp가 telomere  
 . Schulz <sup>18)</sup>  
 telomere 가 ,  
 telomere 가 telomere  
 가 . WRNp가 telomere  
 telomerase  
 telomere  
<sup>12)</sup>  
 WRNp telomere  
 가 가

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## **The Musculoskeletal Tumors of Werner's Syndrome**

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Werner's syndrome is a rare autosomal recessive disorder manifesting as premature aging. It is also known to be characterized by a high frequency of malignant tumors, especially sarcomas. However, Werner's syndrome may be not only a premature aging disease but also a cancer syndrome, because the malignant tumors in these patients are different from those of normal population with respect to involved site, histological type, and age of onset. Recent studies found Werner's syndrome was caused by a mutation of Werner helicase suggesting that WRN helicase may participate in metabolism and repair of DNA. And a dysfunction of WRN helicase may induce the genomic instability causing somatic mutations. Further studies of Werner's syndrome associated with sarcoma might give much informations about the normal aging process and the pathogenesis of sarcomas.

**Key Words :** Werner's syndrome, Premature aging, Sarcoma

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