DIAGNOSTIC CRITERIA FOR WERNER SYNDROME

Last Name________________________ First___________________ Middle________________
Referring Physician________________________________________

Check cardinal signs and symptoms (onset over 10 years old)
   ____ 1. Cataracts (bilateral)
   ____ 2. Characteristic dermatological pathology (tight skin, atrophic skin, pigmentary
   alterations, ulceration, hyperkeratosis, regional subcutaneous atrophy) and
   characteristic facies ('bird' facies)
   ____ 3. Short stature
   ** 4. Parental consanguinity (3d cousin or greater) or affected sibling.
   ____ 5. Premature greying and/or thinning of scalp hair.
   ____ 6. Positive 24-hour urinary hyaluronic acid test, when available.)

Check further signs and symptoms
   ____ 1. Diabetes mellitus.
   ____ 2. Hypogonadism (secondary sexual underdevelopment, diminished fertility, testicular or
   ovarian atrophy).
   ____ 3. Osteoporosis.
   ____ 4. Osteosclerosis of distal phalanges of fingers and/or toes (x-ray diagnosis)
   ____ 5. Soft tissue calcification.
   ____ 6. Evidence of premature atherosclerosis (e.g. history of myocardial infarction).
   ____ 7. Mesenchymal neoplasms, rare neoplasms or multiple neoplasms.
   ____ 8. Voice changes (high pitched, squeaky or hoarse voice).
   ____ 9. Flat feet.

Definite: All the cardinal signs and two others.
Probable: The first three cardinal signs and any two others.
Possible: Either cataracts or dermatological alterations and any four others.
Exclusion: Onset of signs and symptoms before adolescence (except stature, since current
data on pre-adolescent growth patterns are inadequate.)

** Type of consanguinity:
Number of affected sibs:

Registry No________________________
Date of sample submission: Yr______Mo______Day_____ Date of last physical exam: Yr______Mo______Day_____ 

Patient Identification (Confidentiality assured) 
Last name____________________First______________Middle______________Phone_____________
Address______________________________________ __________________________________________
Date of birth Yr____Mo.____Day____ Age at time of this report______
Birth Place City____________________ State or Providence____________________Country_____________
Sex_____ Marital Status_____________Occupation____________________________________________

Attending physician: Name______________________________________  Phone______________________
Address_______________________________________ ______________  FAX_______________________
Contact Person: Name_________________________________________  Phone_____________________
Address_______________________________________ _______________  FAX_______________________
Has this patient been reported in the literature? ________ Cite reference: _______________________ 

Presenting Complaint: ____________________________________________

Family and Social History 
Ethnic background: __________________________
Parental consanguinity?________ Type? (e.g. 1st cousin marriage) ____________________________
Siblings with Werner syndrome? ______________ Please complete a new form for all affected family members
How many children?
Father's Height: _____________  Mother's Height: _____________
Smoking? ___________If yes, _______ packs per day for _______ years

Physical Appearance (Send photos: digital or hard copy) 
Overall appearance of premature aging?
Age at which premature aging began?
Height_________(cm) Weight__________(kilograms) Weight gain/loss?_______ Short Stature? _______
Thin limbs?
Pinched or bird-like facial features?
Loss of hair color?___ Began at what age? _______
Loss of hair?_________ scalp_________ other __________________

Birth and Development 
Born at how many weeks gestation? ________
Birth length _____________(cm) Birth weight___________(kilograms)
Pediatric and adolescent growth history (attach growth charts, if available)__________
Learning disability or mental retardation? ___________
Health problems or surgeries in infancy or childhood?

Eyes 
Were eyes examined by a slit lamp?
Cataracts? Left_______ Right_______ Bilateral_______ Age @ diagnosis______ Age @ surgery_______
Presbyopia?________

Skin and Subcutaneous Tissue (give distribution) 
Change in fat distribution?_______ Regional alterations in amounts of subcutaneous fat?
Tight skin? ___________ Atrophic skin? ___________ Hypermelanosis? ___________
Hyperkeratosis? __________ Soles of feet ___________ Other________________
Ulcerations? (give localization)____________________________
Nail deformity? ___________ Telangiectasia? ___________ Cold Fingers? ___________

Registry No._______________
Oral Cavity/ear/neck/larynx
High pitched, squeaky or horse voice? (describe)_____________
Irregular teeth? __________ Laryngeal atrophy? ____________
Hearing loss? __________ Diminished smell? _____________
Thyroid enlargement? __________ Hypothyroid? __________

Genitourinary
Age of menarche____________ Age of menopause___________ Amenorrhea? _________
Secondary sexual underdevelopment? ______ Atrophic testes?_______
Infertility? __________ Prostate hypertrophy? ____________

Bones, joints, muscles
Flat feet? ______________ Soft tissue calcification? ____________
Osteosclerosis of phalanges of fingers ________ toes________?
Osteoarthritis?
Muscle wasting?
Osteoporosis? __________ Osteopenia? __________ Kyphosis? __________

Cardiovascular
Murmurs? ______________
Evidence of atherosclerosis/arteriosclerosis? ________________
Peripheral vascular disease _________________________________
Coronary artery disease ___________________________________
Medial calcinosis_________________________________________
Hypertension? _________ Blood pressure __________

Neoplasms (benign and malignant)_________________________
(Give histopathologic diagnosis when available. Give age at diagnoses.)

Neurological/Psychological Endocrine
Hyperreflexia? ______________ Clinical dx of diabetes? __________
Mental disorders? _____________ Type of diabetes ______________
Cortical atrophy? ____________ Other _________________________
Dementia? __________ Ataxia? ______________
Neuropathy? __________ Other _________________________

Please attach diagnostic imaging reports (X-ray, MRI, CT, echocardiogram etc) if available

Laboratory medicine results:
Please attach all lab testing, including blood glucose, triglycerides, cholesterol, LDL/HDL, liver function tests, ANA, karyotype or array CGH, any other genetic test results.
24-hour urinary hyaluronic acid levels measured? __________

Pathology results (Biopsy or Autopsy):

Please note any other clinical abnormalities and INCLUDE PHOTOS IF AVAILABLE:

Other valuable documents would include clinic notes by medical geneticists, discharge hospital summaries, anatomic pathology results and clinical lab reports, x-ray reports of any significant findings, and autopsy reports of siblings.
Would this patient and patient’s next-of-kin be likely to agree to autopsy examination? yes_______ no________