INTERNATIONAL REGISTRY OF WERNER SYNDROME AND OTHER PROGEROID SYNDROMES

UNIVERSITY OF WASHINGTON, DEPARTMENT OF PATHOLOGY
BOX 357470, SEATTLE, WA 98195-7470 USA
Phone: (206) 543-5088 Fax: (206) 685-8356

******PLEASE COMPLETE ALL THREE PAGES AND SEND WITH SAMPLE******

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_____

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PHONE: (206) 543-5088 Fax: (206) 685-8356 Website: wernersyndrome.org

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? _____

Name_____ First_____ Middle_____

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

Hyperreflexia? _____
Mental disorders? _____
Cortical atrophy? _____
Dementia? _____ Ataxia? _____
Neuropathy? _____ Other _____

Endocrine

Clinical dx of diabetes? _____
Type of diabetes _____
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 AVAILALE:

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_____