ERN-SKIN / ERRAS DNA Albinism spec Dataset

1. Pseudonym

1.1 SPIDER ID (Individual ID):

1.2 Alternative ID:

2. Personal information

2.1 Date of Birth

2.2 Sex at birth
- ☐ Female
- ☐ Intersex
- ☐ Male
- ☐ Undetermined

2.3 Country of Birth

3. Patient Status

3.1 Patient Status
- ☐ Alive
- ☐ Dead   **3.2 Date of death:**
- ☐ Lost in follow-up
- ☐ Opted-out
- ☐ Unknown

4. Care pathway

4.1 Date first contact with specialised center

4.2 Healthcare Provider   Necker

4.3 Physician's name

5. Disease history

Age at onset	Onset Date:	

6.1 Diagnosis of the rare disease

6.1.1 Rare disease classification (ORPHA)	Code ORPHA:	
6.1.2 Rare disease classification (ICD10)	Code ICD10:	

6.2 Genetic Diagnosis

6.2.1 Gene	List of Genes	

6.2.2 Chromosome Analysis Test performed?	☐ Yes			
	☐ No			
6.2.3 Genetic tests performed?	☐ **Yes:**	**Date testing yyyy/mm**	
	☐ No	**Mutated gene and mutation**	
	☐ Unknown		
6.2.4 DNA analysis performed?	☐ **Yes, DNA technique:**	☐ **Sanger**	
	☐ No	**NGS targeted panel**	
		WES open	
		WES panel	
		WGS	
		☐ **Other:**	
		Specify other DNA technique	

6.3 Histologic Diagnosis

6.3.1 Standard histology	☐	Yes	
	☐	No	
6.3.2 Antigen mapping	☐	Yes, result:	
	☐	No	
6.3.3 Electron microscopy	☐	Yes	
	☐	No	

6.4 Immunology Diagnosis

6.4.1 Antibody results	
6.4.2 Name Ab + name company	
6.4.3 Target Protein	

6.5 Undiagnosed case

6.5.1 Clinical symptoms and physical findings in patient (HPO: Human Phenotype Ontology)	
6.5.2 Negative phenotype annotations (HPO: Human Phenotype Ontology)	

6.6 Etiology

6.6.1 Drug etiology: which drug?	
6.6.2 Infectious etiology: which infection?	
6.6.3 other?	
6.6.4 Unknown Etiology?	

7. Research/consent

7.1 Agreement to be contacted for research purposes or clinical trials	☐ Yes ☐ No
7.2 Consent to the reuse of data	☐ Yes ☐ No
7.3 Patient's biological sample available for research	☐ Yes ☐ No
7.4 Biological sample stored in a biobank	☐ **Yes** ☐ No
7.5 Link to a Biobank:	

8.Disability

8.1 Classification of functioning/disability (ICF)	

9. Images available

9.1 Clinical photographs available? (ex: photo's of the patient's skin)		Yes			
Photos are available and can be requested to be shared later.		No			
9.2 Medical images available? (ex: radiology)		**Yes:**		radiography	
Images are available and can be requested to be shared later.		No		scanner	
				engiography	
				other:	
9.3 Histology image available? (microscopic image)		**Yes:**			
Images are available and can be requested to be shared later.		No			
9.3.1 Date Histology image					
9.3.2 Comments histology image					
9.4 Extra comments on images					

Demographic data

Education	Yes
	No
Professional activity	Yes
	No
If answer yes, please provide the profession	
Parents consanguinity	Yes
	No
If answer yes, please explain	
Affected sibling	**Yes:**
	Brother
	Sister
	No
Ethnicity	

Medical and paramedical follow up

Name reference centre Follow up:	
Dermatologist Follow up:	
visit(s)/year	
Title	
First name	
Last name	
Name reference centre Follow up:	
Ophthalmologist Follow up:	
visit(s)/year	
Title	
First name	
Last name	
Name reference centre Follow up:	
General practitioner Follow up:	
visit(s)/year	
Title	
First name	
Last name	

Medical history

Skin color at birth:	Pale white skin		
	Fair skin		
	Darker white skin		
	Light brown skin		
	Brown skin		
Hair color at birth:	Platinium		
	White		
	Blond		
	Brown		
	Dark brown		
	Red hair		
Skin cancers:	No		
	Yes, complete below:		
Age at onset (year + month):			
Basocellular Carcinoma	No		
	Yes, complete :	Unique	
		Multiple	
		Date (YYYYMM):	
		Location:	
Spinocellular Carcinoma	No		
	Yes, complete :	Unique	
		Multiple	
		Date (YYYYMM):	
		Location:	
Pre-epithelial keratosis	No		
	Yes, complete :	Unique	
		Multiple	
		Date (YYYYMM):	
		Location:	
Melanoma	No		
	Yes, complete :	Unique	
		Multiple	
		Date (YYYYMM):	
		Location:	

Medical history

	Condition/Illness/surgical procedure/cancer	Start date (YYYY/MM/DD)	Stop date (YYYY/MM/DD)	Or tick if ongoing at Visit?	Related to XP	Medication (If yes, complete medication table)
1				☐	☐ Yes ☐ No	☐ Yes ☐ No
2				☐	☐ Yes ☐ No	☐ Yes ☐ No
3				☐	☐ Yes ☐ No	☐ Yes ☐ No
4				☐	☐ Yes ☐ No	☐ Yes ☐ No
5				☐	☐ Yes ☐ No	☐ Yes ☐ No
....				☐	☐ Yes ☐ No	☐ Yes ☐ No
Possibility to add lines						

Medication

	Medical history N°	Medication (Record Generic or trade name)	Dose and unit	Frequency	Routs	Start date (YYYY/MM/DD)	Stop date (YYYY/MM/DD)	Or tick if ongoing at Visit?
1								☐
2								☐
3								☐
4								☐
5								☐
....								☐
Possibility to add lines								

Family medical history

	Condition/Illness/surgical procedure/cancer	Start date (YYYY/MM/DD)	Stop date (YYYY/MM/DD)	Or tick if ongoing at Visit?
1				☐
2				☐
3				☐
4				☐
5				☐
....				☐
Possibility to add lines				

Patient's Fitzpatrick Skin Type	No		
	Yes, complete :	I Burns easily, rarely tans	
		II Burns easily, tans minimally	
		III Burns moderately, tans gradually	
		IV Burns minimally, tans well	
		V Rarely burns, tans profusely	
		VI Never burns, deeply pigmented	
Mother's Fitzpatrick Skin Type	No		
	Yes, complete :	I Burns easily, rarely tans	
		II Burns easily, tans minimally	
		III Burns moderately, tans gradually	
		IV Burns minimally, tans well	
		V Rarely burns, tans profusely	
		VI Never burns, deeply pigmented	
Father's Fitzpatrick Skin Type	No		
	Yes, complete :	I Burns easily, rarely tans	
		II Burns easily, tans minimally	
		III Burns moderately, tans gradually	
		IV Burns minimally, tans well	
		V Rarely burns, tans profusely	
		VI Never burns, deeply pigmented	
Clinical data	weight (kgs)		
	height (meter)		

Skin symptoms

No			
Yes, complete:	Homogeneous depigmentation	no	
		yes	
	Color of skin:	Creamy	
		White	
		Pink	
		Brown	
	Color of eyelashes:	white	
		light blond	
		dark blond	
		red	
	Color of scalp hair:	white	
		light blond	
		dark blond	
		red	
	Color of eyebrows:	white	
		light blond	
		dark blond	
		red	
	Color of body hair:	white	
		light blond	
		dark blond	
		red	
	Tanning	no	
		yes	
	Nævus	no	
		yes: age	YYMM
	Telangiectasia	no	
		yes: age	YYMM
	Actinic lesions	no	
		yes: age	YYMM

Skin cancers:

Basal cell carcinoma	
Squamous cell carcinoma	
Melanoma	
Other Skin Cancer	

Ophthalmological Data:

Ophthalmological involvement:	No	☐
	Yes, complete below:	☐
Photophobia	Yes	☐
	No	☐
Low visual acuity	Yes	☐
	No	☐
Nystagmus	Yes	☐
	No	☐
Hypermetropia	Yes	☐
	No	☐
Myopia	Yes	☐
	No	☐
Strabismus	Yes	☐
	No	☐
color of eye (iris):	grey	☐
	blue	☐
	green	☐
	brown	☐
Slit-lamp examination:		
Iris transillumination	No	☐
	Yes, complete below:	☐
	Grade 1	☐
	Grade 2	☐
	Grade 3	☐
	Grade 4	☐
Foveal hypoplasia	No	☐
	Yes, complete below:	☐
	Grade 1	☐
	Grade 2	☐
	Grade 3	☐
	Grade 4	☐
Abnormal routing of fibers Visual Evocated Potential	Yes	☐
	No	☐

Neurological Data

Neurological involvement	Yes	☐
	No	☐
Neurodevelopmental delay	Yes	☐
	No	☐

Hematological Data

Hematological Symptoms	No	☐
	Yes, complete Hematological Type:	☐
	Bleeding	☐
	Platete secretion test	☐

Digestive symptoms (abdominal pain, diarrhea)

	Yes	☐
	No	☐

Pulmonary fibrosis

Pulmonary fibrosis	No	☐
	Yes	☐
Other		

Photoprotection

Sunglasses	Yes	☐
	No	☐
Close Covering Clothes	Yes	☐
	No	☐
Hat	Yes	☐
	No	☐
Parasol	Yes	☐
	No	☐
Suit	Yes	☐
	No	☐
Is the patient using sunscreen cream?	No	☐
	Yes, complete below:	
Trade name:		
Used on Skin:	Yes	☐
	No	☐
Used on Lips:	Yes	☐
	No	☐

Genetics data

Molecular analysis performed	Yes	☐
	No	☐
Panel NGS	Yes	☐
	No	☐
Other technics: WGS	Yes	☐
	No	☐
Mutated gene:	CHS1	☐
	HPS1	☐
	HPS2	☐
	HPS3	☐
	HPS4	☐
	HPS5	☐
	HPS6	☐
	HPS7	☐
	HPS8	☐
	HPS9	☐
	HPS10	☐
	HPS11	☐
	OCA1	☐
	OCA2	☐
	OCA3	☐
	OCA4	☐
	OCA5	☐
	OCA6	☐
	OCA7	☐
	OCA8	☐
	Unkown	☐
Mutations:		
Sporadic	Yes	☐
	No	☐
Familial	Yes	☐
	No	☐
Mode of inheritance:	X linked	☐
	Autosomal / recessive	☐
	Autosomal detruiment	☐

Environmental data

What is the means of transport frequently used by the patient?	Family car		
	Taxi		
	School bus		
	Public transport		
	Other:		
Physical activity	Kind of activity:		
	Mean frequency:		/week
			/month
Does the patient have leisure time activities?	No		
	Yes, complete below:		
	Kind of activity:		
	Mean frequency:		/week
			/month