

# The care as perceived by individuals with Shwachman Diamond Syndrome (SDS) in Canada, Italy, Netherlands, United Kingdom and United States.

Liesbeth Siderius , Shwachman Diamond Syndrome Support Holland, the Netherlands

On behalf of Shwachman Diamond Syndrome "United": Shwachman Diamond Syndrome Canada Inc, Canada; Shwachman Syndrome Italian Association, Italy; Shwachman Diamond Syndrome Support , Netherlands; Shwachman Diamond Syndrome Foundation, UK; Shwachman-Diamond Syndrome Foundation, USA

Shwachman Diamond Syndrome (SDS) was described in 1964 on the basis of clinical manifestations: bone marrow failure, a pancreas insufficiency, and short stature. This multisystem autosomal recessive disorder is caused by a gene, discovered in 2003. SDS has an estimated frequency of 1:100.000-150.000. A genetic diagnosis of SDS, makes coordinated – preventive - care feasible. We questioned the persons with SDS in 5 different countries on the care as perceived.

Response:

	Canada	Italy	NL	UK	US	BE	Total
population	33000000	60000000	16000000	60000000	306000000	10000000	
SDS estimated	220	400	107	400	2040	67	3234
SDS registered	31	57	15	55	144		
regis % estim	14,1	14,2	14	14	7		
SDS response	4	31	11	5	11	2	<b>64</b>
% totaal estim.	1,8	7,75	10,2	1,25	0,5	2,9	
% part of study	6,3	48,4	17,2	17,2	7,8	3,1	

Population of country; Persons with SDS when frequency = 1:150.000; Persons registered by the national patient organization; % of registered persons/ estimated persons with SDS; respondents on request of the national patient organization; % responders per estimated persons with SDS; % individuals per country as part of this study. The Belgian families are registered in the Dutch foundation

The **Italians** receive **coordinated** care **significantly** more frequent (p=.040) as compared to the whole group , more specific as compared to persons in the Netherlands and the United States.

	Can	Ita	NL	UK	US	Be	Total
Coordinat	3	20	3	2	3	2	33
total	4	29	10	5	11	2	61
	75%	69%	30%	40%	27%	100%	51%

73% consult a **gastroenterologist** , less then half of these are perceived to have expertise in SDS.

86% consult a **hematologist**, half of these are perceived to have expertise in SDS.

16 % of al individuals with SDS are visiting **bone / skeletal** specialists. 54% has had skeletal X ray's. This may mean that either the skeletal system is not involved or the skeletal abnormalities are surveyed by other care takers. The diagnosis SDS may not be considered in individuals with major skeletal abnormalities.

81% of the persons visit a (local) **dentist**, 11% visited a dentist with expertise on SDS.

39 % of individuals of 16 years and older experience difficulties in **finding employment**. Of all individuals 31 % gets **psychological** support.

In 81 % of the individuals **DNA studies** were performed.

More expertise on each manifestation of SDS , identification of possibly unrecognized features and coordination and collaboration between health care providers , as wished by the majority of responders, seems justified by this survey.

Needed ! : **International accepted guidelines on diagnosis and care and an international data registration**

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