

Supporting Shwachman Diamond Syndrome patients and families with a multifunctional portal

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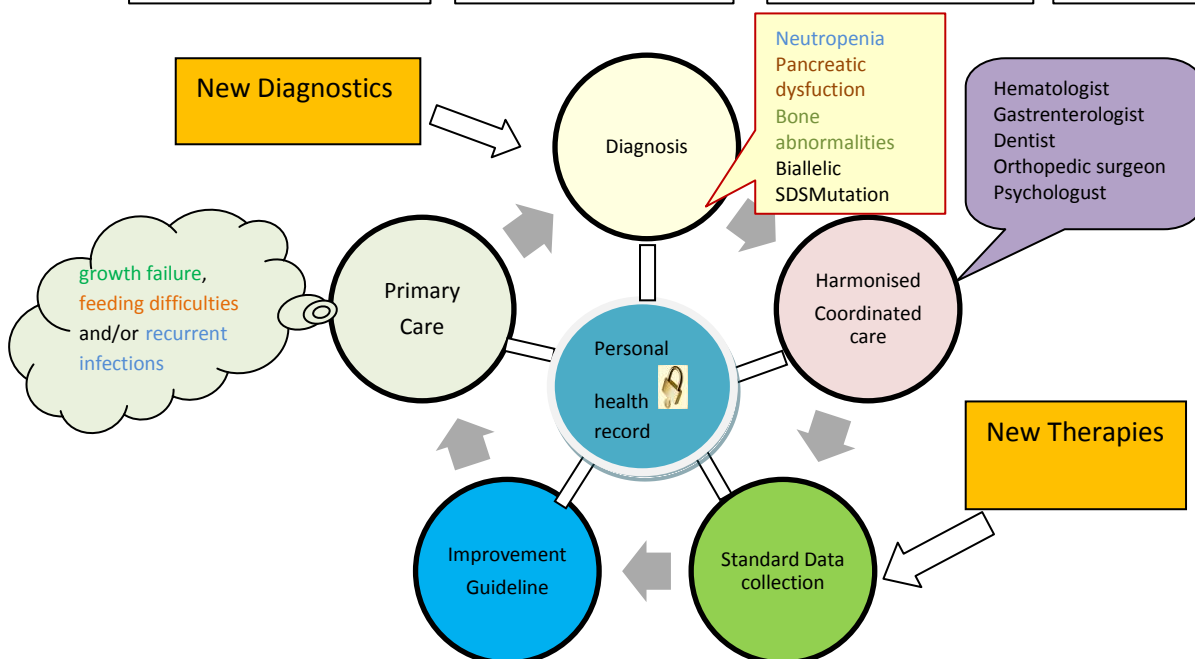
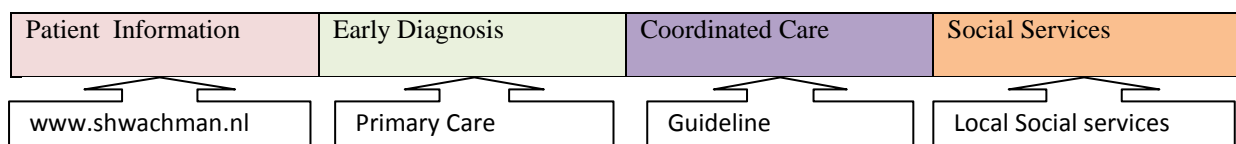
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Rare diseases are generally poorly understood and little information about the **natural history** or management is available. European countries differ in population size, economy and health care systems. In 2009 a survey was undertaken to discover the needs of individuals with Shwachman Diamond Syndrome (SDS). They wish more expertise on **recognition** of feature of SDS, and **coordination of care** according to an international guideline. Technology promotes consistency in the application of guidelines. Evidence outcomes is sparse and would be welcomed by both patients and care-providers.

Integrated into a **portal** for SDS, and potentially for other rare diseases.

The functions include

- resources on symptoms, diagnosis, treatment, care, and quality of life.
- peer support, and the “yellow pages” for health and social services are important functions.



A **personal health record** (PHR), the patient can document : symptoms, test results, and treatment

- **control of access** of providers to personal health data, and
- **exchanging** information with professional electronic health records.
- **opt in** with research studies and contribute to a clinical data warehouse to improve long term outcome .