



eb Clinet Conference 2024

Transition from Paediatric to Adult Care
17th October 2024

Dr Anna Martinez and Janet Hanson



NHS

**Great Ormond Street
Hospital for Children**
NHS Foundation Trust

NHS

Guy's and St Thomas'
NHS Foundation Trust

What we will cover

- Physiology of puberty & how having severe types of EB can effect this
- Key publications on EB transition
- How we plan for transition & some useful resources to use
- A video showing 1 young person with EB who has now transferred her care to the adult services with their insightful & powerful feedback
- Questions and Answers

Causes of endocrine & growth disturbance in severe EB

Multifactorial

- nutritional deficiencies
- drugs: steroids mostly but we rarely use in EB

Chronic inflammation

Wounds & tissue damage or infection triggers nucleated cells to produce cytokines

Most important cytokines are:

interleukin 6 (IL-6)

interleukin 1 (IL-1)

tumour necrosis factor alpha (TNF α)



Received: 15 October 2023 | Revised: 17 January 2024 | Accepted: 10 February 2024

DOI: 10.1111/exd.15040

RESEARCH ARTICLE

Experimental Dermatology | WILEY

Elevated expression of interleukin-6 (IL-6) and serum amyloid A (SAA) in the skin and the serum of recessive dystrophic epidermolysis bullosa: Skin as a possible source of IL-6 through Toll-like receptor ligands and SAA

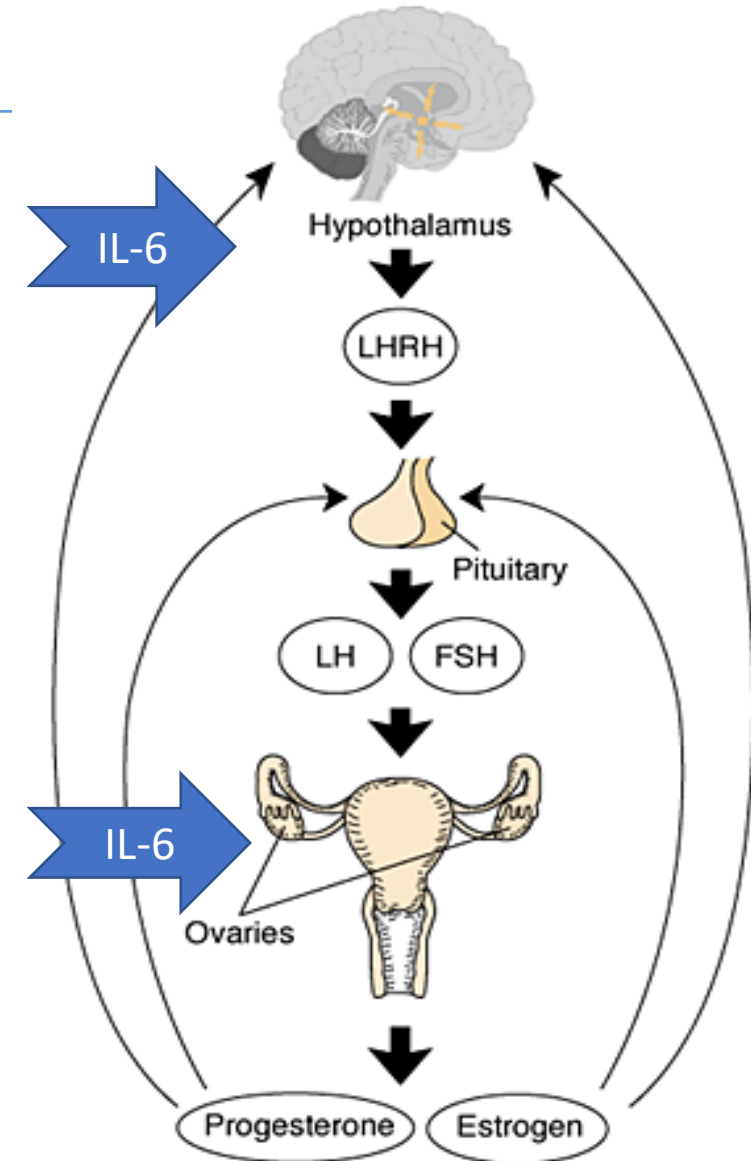
Yoshio Kawakami  | Ai Kajita | Ken-Ichi Hasui | Yoshihiro Matsuda | Keiji Iwatsuki | Shin Morizane 

Affects of cytokines on body pathways

Hypothalamic-pit-gonadal axis	➡	pubertal delay
GH/IGF-1 axis	➡	growth delay
Bone regulatory pathway	➡	osteoporosis & fractures
Iron metabolism	➡	anaemia

Hypothalamic-pituitary- gonadal axis

Secondary
hypogonadotropic
hypogonadism



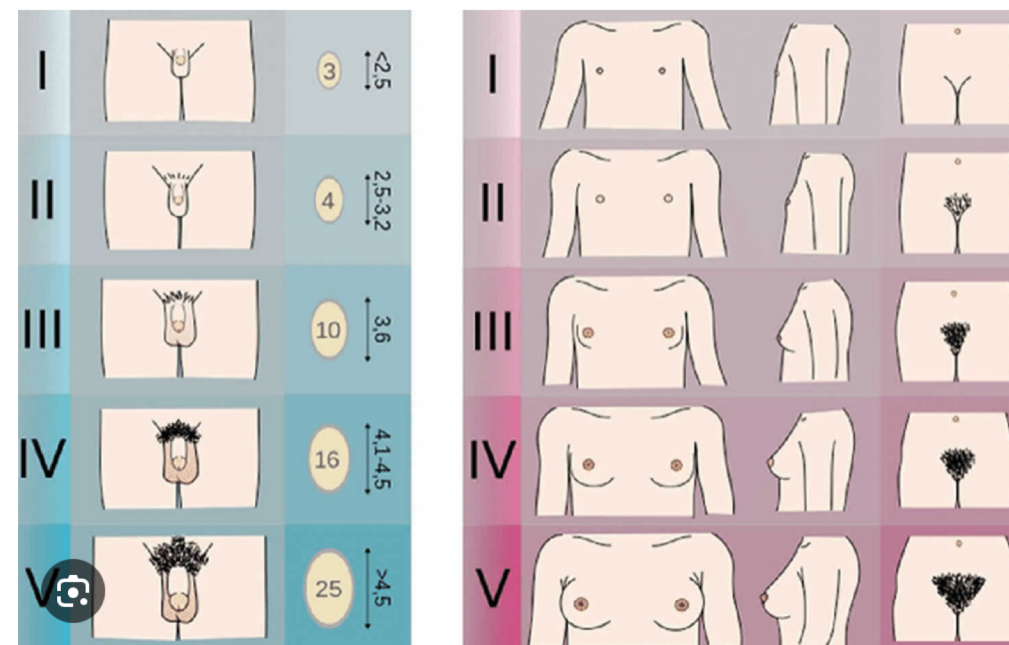
Definition of pubertal delay

Girls

- No breast development by 13 years or no menarche by 16 yrs

Boys

- Testicular volume <4mls over 14 yrs



Tanner puberty scale for male and female

[Visit >](#)

How common is pubertal delay in EB?

> [Pediatr Dermatol.](#) 2023 Jan;40(1):100-106. doi: 10.1111/pde.15136. Epub 2022 Sep 25.

Prevalence of delayed puberty and low bone density in patients with epidermolysis bullosa: Insight from a large single center's experience

Halley Wasserman^{1 2}, Andrea Dumenigo³, Lindsey Hornung¹, Bret Augsburg¹,
Kalyani Marathe^{1 2}, Anne W Lucky^{1 2}

Retrospective review Jan 2010-Sept 2022
total of 186 individuals with different types of EB

98 with RDEB

33% pubertal delay
29% low BMD by age 10 years

In the other subtypes pubertal delay was inconclusive

[PLoS One.](#) 2022; 17(9): e0274072.

Published online 2022 Sep 6. doi: [10.1371/journal.pone.0274072](https://doi.org/10.1371/journal.pone.0274072)

PMCID: PMC9447886

PMID: [36067143](https://pubmed.ncbi.nlm.nih.gov/36067143/)

A single-centre study on predictors and determinants of pubertal delay and growth impairment in Epidermolysis Bullosa

[Giulia Rodari](#), Conceptualization, Data curation, Formal analysis, Project administration, Software, Writing – original draft, Writing – review & editing,^{1, 2, *} [Sophie Guez](#), Data curation, Investigation, Writing – review & editing,³ [Simona Salera](#), Investigation, Writing – review & editing,³ [Fabio Massimo Olivieri](#), Data curation, Investigation, Software, Writing – review & editing,⁴ [Gianluca Tadini](#), Data curation, Investigation, Writing – review & editing,⁵ [Michela Brena](#), Investigation, Supervision,⁵ [Eriselda Profka](#), Supervision, Writing – review & editing,¹ [Federico Giacchetti](#), Conceptualization, Writing – review & editing,¹ [Maura Arosio](#), Validation, Writing – review & editing,^{1, 2} and [Claudia Giavoli](#), Supervision, Validation, Writing – review & editing^{1, 2}

21 patients- mean age 16.2+/- 2.5 years

12 RDEB/ 3 DDEB

2 EBS

3 JEB

1 kindler EB

Results

Pubertal delay 7/21 -33% (6 RDEB & 1 JEB)

Low BMD in RDEB 50%

Expect pubertal delay in severe types of EB up to **third** of individuals

- Need to address both medical & psychological aspect of pubertal delay
- 40 % bone mass accrued during puberty so enormous impact on bone health

Pubertal attainment crucial

Endocrinologist
Psychologist



The important of Sexual Health

Promoting sexual health & wellbeing

- Address sexual health care as for general population
- Specific to the needs of individuals with EB

King et al. *Orphanet J Rare Dis* (2021) 16:9
<https://doi.org/10.1186/s13023-020-01640-0>


Orphanet Journal of
Rare Diseases

REVIEW

Open Access

Supporting sexuality for people living with epidermolysis bullosa: clinical practice guidelines





Alex King^{1*} , Humphrey Hanley², Mark Popenhagen¹, Florencia Perez³, Kerry Thompson⁴, Diana Purvis⁵, Nora Garcia⁶, Ida Steinlein⁷, Mia Werkentoft⁸, Matthew Lightfoot⁹, Michelle Lahat¹⁰, Kalsoom Begum¹¹ and Julio Tanabe¹²

Transition in EB literature- 5 articles

Received: 6 September 2022 | Accepted: 9 December 2022
DOI: 10.1111/pde.15232

Pediatric
Dermatology WILEY




Transition of care in patients with epidermolysis bullosa: A survey study

Joohee Han MD¹ | Morgan Dykman BA¹  | Scott Lunos MS² |
Audrey Nguyen¹ | Kristen P. Hook MD¹ | Irene Lara-Corrales MD³  |
Harper N. Price MD⁴ | Lucia Diaz MD⁵ | Moise Levy MD⁵ | Christina Boull MD¹

Received: 10 March 2023 | Accepted: 2 April 2023
DOI: 10.1111/pde.15330

Pediatric
Dermatology WILEY

Transition readiness in adolescents and young adults with chronic genetic skin conditions

Frances O. Ho BA¹  | Andrea M. Rustad BA¹  | Ziyu Ren PhD¹ |
Lydia R. Khabbaz PhD¹ | Justin M. Knoll PhD² | Stephanie M. Rangel PhD¹ |
Amy S. Paller MD¹ 

Review of transition of care literature: Epidermolysis bullosa—A paradigm for patients with complex dermatologic conditions



Victoria A. Perez, BA, MS,^a Kimberly D. Morel, MD,^{b,c} Maria C. Garzon, MD,^{b,c}
Christine T. Lauren, MD, MHA,^{b,c} and Laura E. Levin, MD^b
New York, New York

J AM ACAD DERMATOL
SEPTEMBER 2022

Puberty and EB

Transition from paediatric to adult service in epidermolysis bullosa



Lesley Foster, Yvette Holmes

British Journal of Nursing, 2007, Vol 16, No 4



Box 1. Summary of transition programme

Coordinator(s) of programme: Paediatric EB Clinical Nurse Specialists (CNS)

Age of transition events

First discussion of transition to Adult EB service may be mentioned as early as the child's diagnosis	Discuss adult centres and transition plan from 13 years
Beginning of active preparation for transition	14–16 years
Transfer of care to adult service	16–18 years

Target adult service used—Adult EB nurses St Thomas' Hospital, London
Heartlands Hospital, Birmingham

Transition programme elements

GOSH staff to encourage independence of young adult by encouraging participation in consultations, eventually leading to self reliance in medical discussions	Agreed programme and timetable Introduction to adult EB clinical nurse specialist team by informal contacts Discussion of relevant topics to increase understanding, e.g.: condition, healthcare system, benefits Visit to one or both adult centres, as required Home visit by an adult CNS Ongoing support by paediatric CNS until transfer Attendance by paediatric CNS at initial adult outpatient consultations
GOSH professionals to inform shared care (hospital and community staff)	

Evaluation procedures

Discussion and review with EB adult CNS
Review of progress with patient and evaluation of transition programme

Review > J Am Acad Dermatol. 2022 Sep;87(3):623-631. doi: 10.1016/j.jaad.2020.06.083.

Epub 2020 Jun 24.

Review of transition of care literature: Epidermolysis bullosa–A paradigm for patients with complex dermatologic conditions

Victoria A Perez ¹, Kimberly D Morel ², Maria C Garzon ², Christine T Lauren ², Laura E Levin ³

Affiliations + expand

PMID: 32592878 DOI: 10.1016/j.jaad.2020.06.083

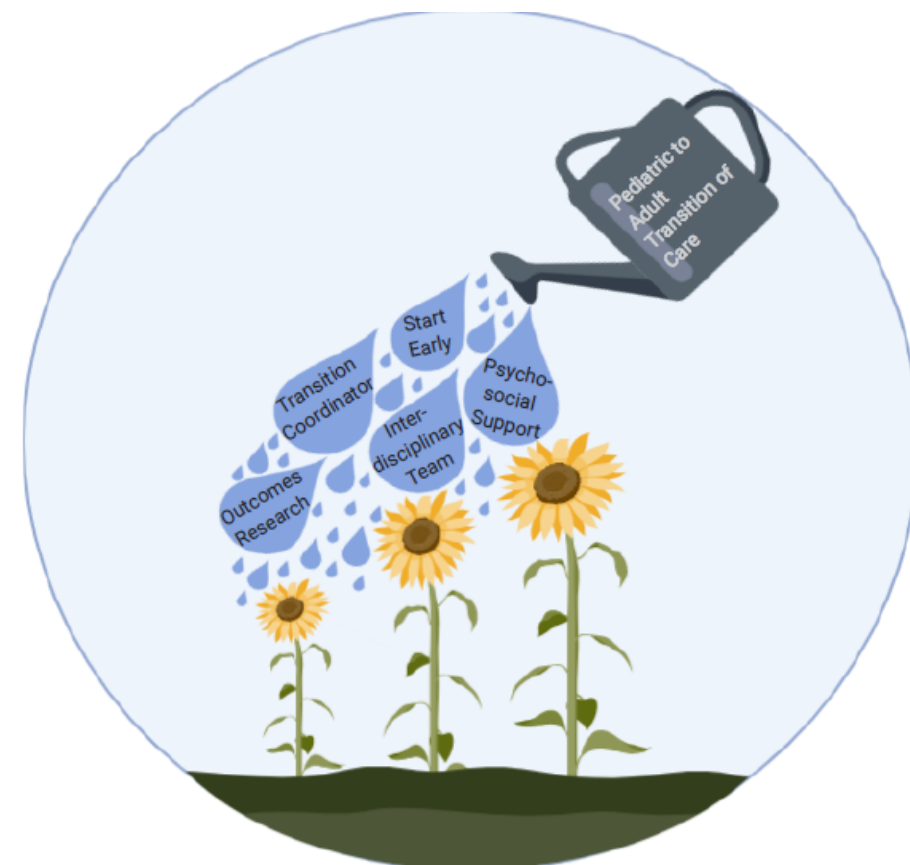
Definitions

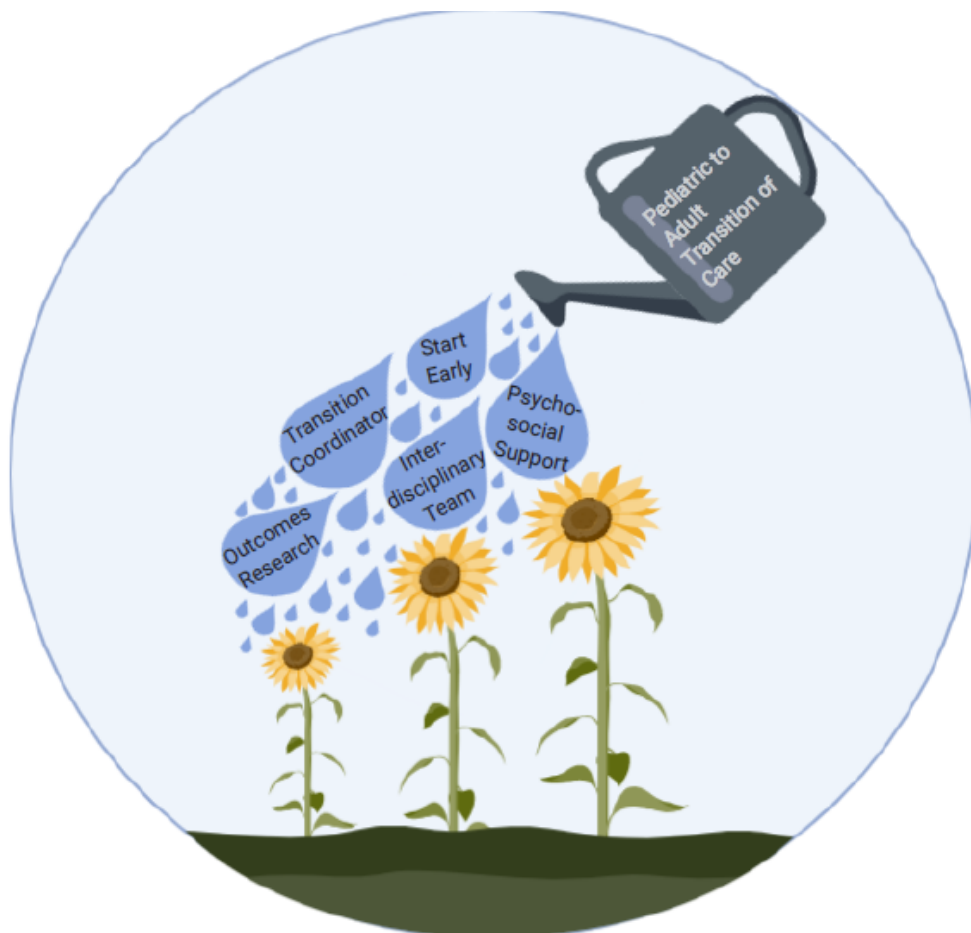
The purposeful planned movement from child-centred care to an adult-oriented health care system

- To improve the ability of young adults to manage their own health care needs transferring to adult health care services

Transition Process

Transfer Final event of handing over care



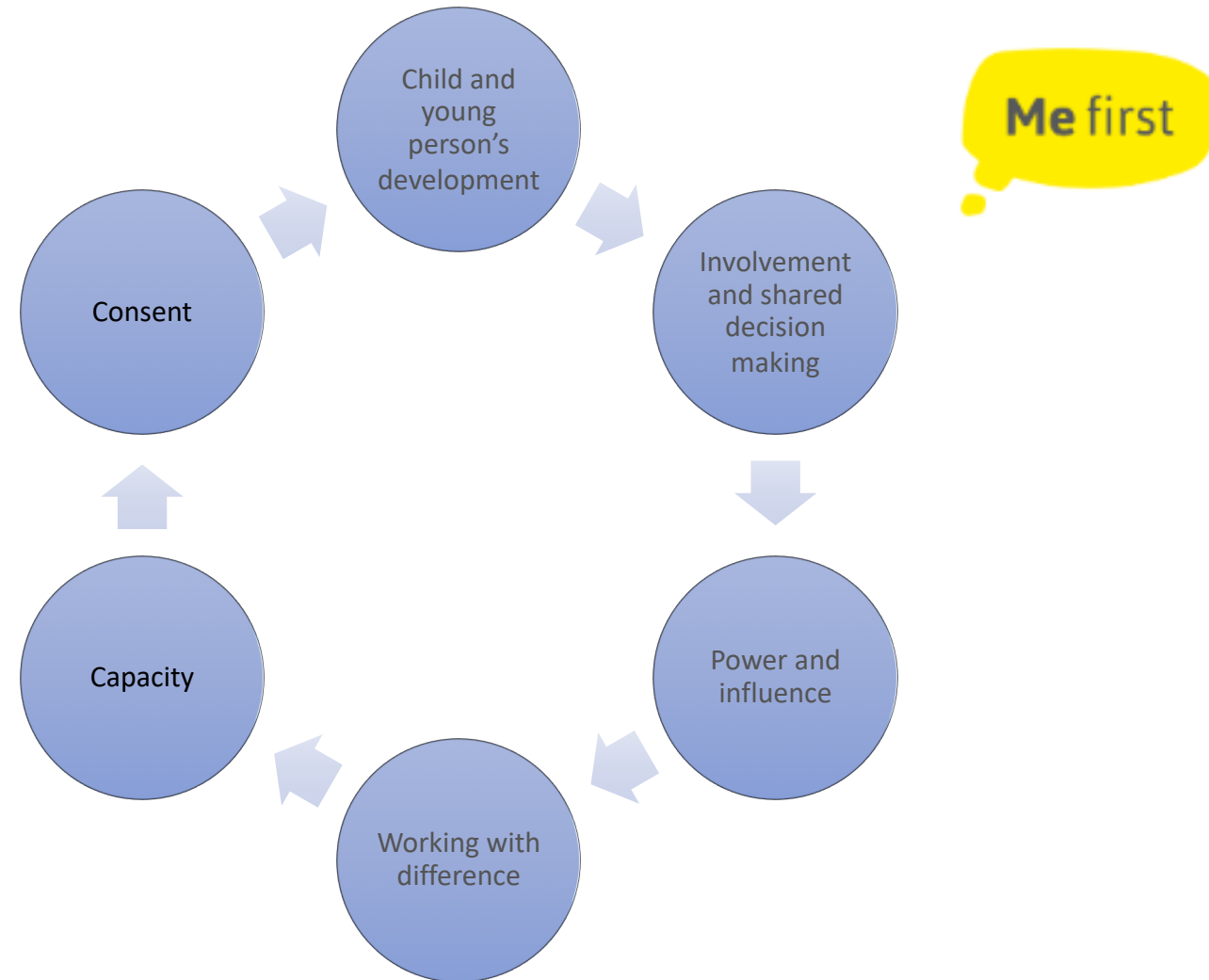


- Literature review – transition
- Only 36 papers relevant
- Start early between 12-14 years of age
- Need an maintain the multidisciplinary team approach and build up de-novo relationships
- Transitioning lead
- Psychology support

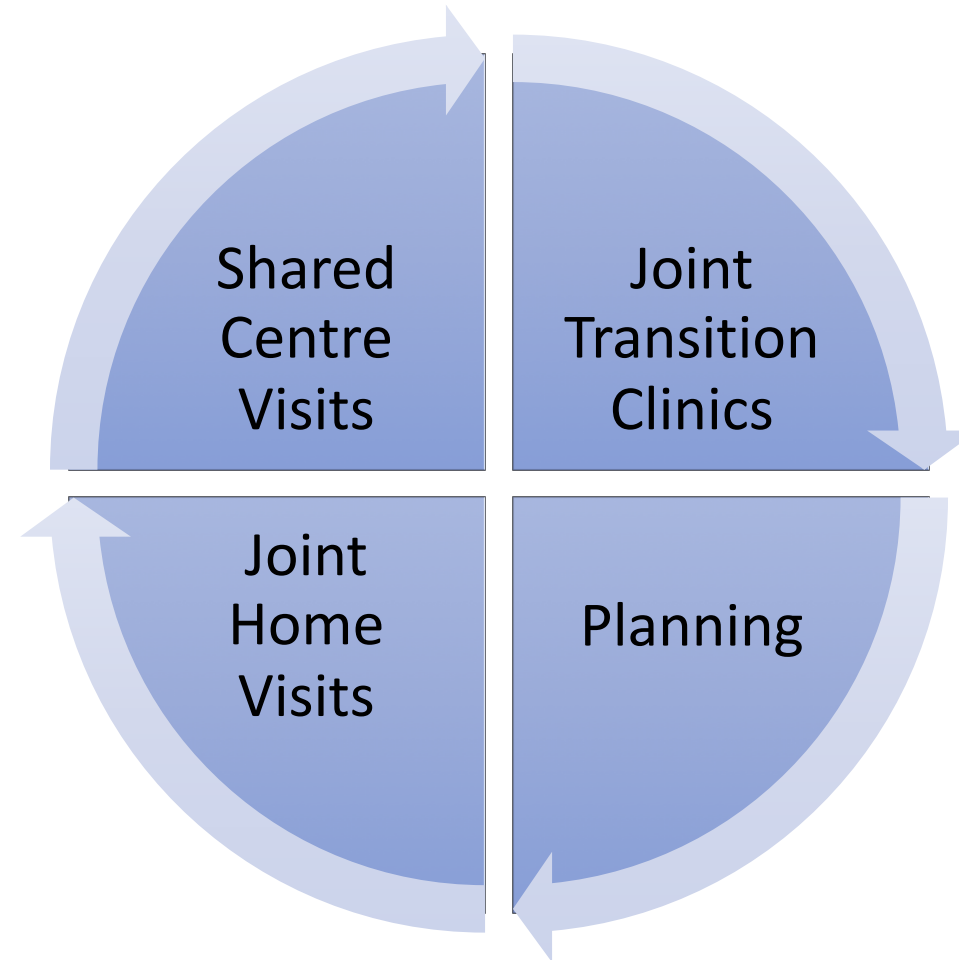
Challenges with EB Transition



Safe and Successful Transition



Transition planning Processes



Ready Steady Go



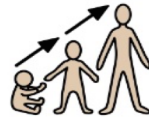
[Ready Steady Go - TIER Network](#)

Growing Up and Gaining independence



Great Ormond Street **NHS**
Hospital for Children
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Growing up, gaining independence: families



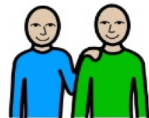
Your child is getting older. One day, they will leave GOSH to go to a different hospital.



We want to help you to get your child ready. We will work with you to help them learn.



We will show you how to talk to your child about their health.



We think it is good for young people to see the doctor without their parents for some of the appointment. A member of staff can go with them if they want.



When your child is 16 years old, the law lets them make choices. We will help work out if they can make decisions on their own. We call this 'having capacity'.



If they can, your child will be the main person to decide about their health.



The hospital should send letters to your child instead of you.



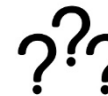
If they can, they will decide who can see the information GOSH keeps about them.



Your child will have lots to learn. You can help them with lots of things.



We know you might feel sad when your child takes charge of their health. This is a normal part of growing up. You can help them get ready.



Please ask us if you have any questions.

Ref: 2017ER0191 August 2018

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Compiled by the Transition Improvement Manager in collaboration with the Child and Family Information Group at Great Ormond Street Hospital for Children NHS Foundation Trust.
Widgit Symbols (c) Widgit Software 2002-2012 (www.widgit-health.com)

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Resources



[Healthcare Transition | Great Ormond Street Hospital \(gosh.nhs.uk\)](https://gosh.nhs.uk)

Podcasts available of Spotify



Acknowledgment & Thanks

